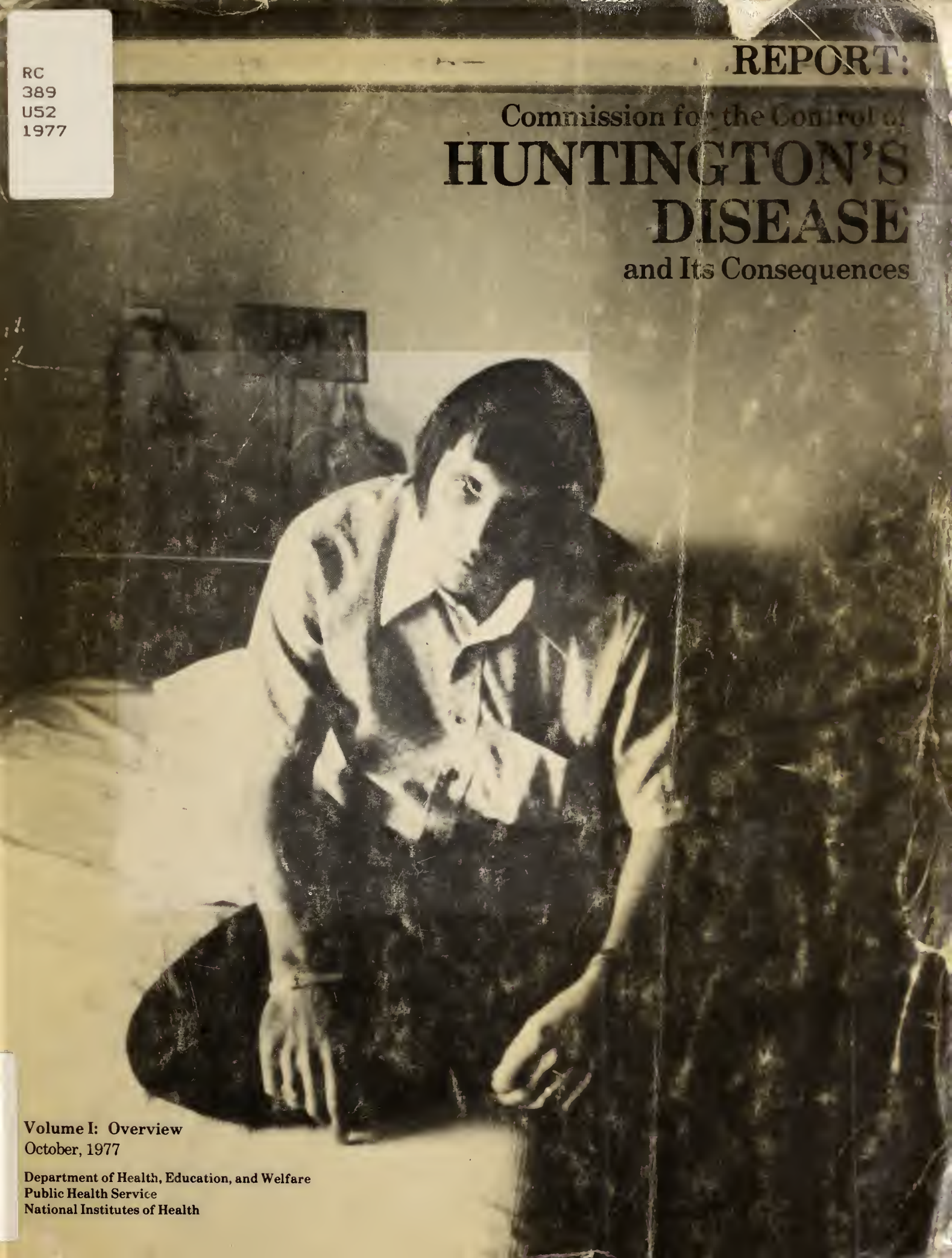
REPORT:

Commission for the Control of HUNTINGTON'S DISEASE and Its Consequences



Volume I: Overview
October, 1977

Department of Health, Education, and Welfare
Public Health Service
National Institutes of Health

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Commission for the Control of
HUNTINGTON'S DISEASE
and Its Consequences

Volume I — Overview

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U.S. Department of Health, Education, and Welfare
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Congressional Mandate

The 94th Congress, through Public Law 94-63, established the Commission for the Control of Huntington's Disease and Its Consequences. The law stipulated that the Secretary of Health, Education, and Welfare appoint nine members to the Commission: six professionals, chosen for their long-standing research or clinical interest in the disease, and three lay persons, chosen for their dedication to health care needs or their familial involvement with Huntington's disease. The Commission was charged:

1. To make a comprehensive study of the state of the art of medical and social management of Huntington's disease in the United States.

2. To investigate and make recommendations concerning the proper roles of federal, state and local governments and national and local public and private agencies in research, prevention, identification, treatment, and rehabilitation of persons with Huntington's disease.

3. To develop a comprehensive national plan for the control of Huntington's disease and its consequences based on the most thorough, complete, and accurate data and information available on the disorder.

4. To transmit to the President and the Congress on August 31, 1977 a report detailing the findings and conclusions of the Commission, together with recommendations for legislation and appropriations.

P.L. 94-63
Section 605(b)

Commission Members

Chairperson

Marjorie Guthrie
Committee to Combat Huntington's Disease
New York, New York

Vice Chairperson

Milton Wexler, Ph.D.
Hereditary Disease Foundation
Los Angeles, California

Commissioners

Stanley M. Aronson, M.D.
Dean of Medicine
Brown University
Providence, Rhode Island

Ching Chun Li, Ph.D.
University Professor
Department of Biostatistics
University of Pittsburgh
Pittsburgh, Pennsylvania

Guy McKhann, M.D.
Professor and Head
Department of Neurology
School of Medicine
The Johns Hopkins University
Baltimore, Maryland

Alice E. Pratt
Huntington's Chorea Foundation
Houston, Texas

Lee E. Schacht, Ph.D.
Supervisor
Human Genetics and Health Resources for the Handicapped Unit
Minnesota Department of Health
Minneapolis, Minnesota

Jennifer Jones Simon
Malibu, California

Stanley Stellar, M.D.
Director
Neural Sciences Research Institute
St. Barnabas Medical Center
Livingston, New Jersey

Commission Staff

Nancy Wexler, Ph.D.
Executive Director

Charles MacKay, Ph.D.
Deputy Director

Elizabeth McDonald
Administrative Assistant

to Woody, Leonore, and Fletcher

DEPARTMENT OF HEALTH, EDUCATION, AND WELFARE

COMMISSION FOR THE CONTROL OF HUNTINGTON'S DISEASE AND ITS CONSEQUENCES
Building 31, Room 8A11 National Institutes of Health Bethesda, Maryland 20014 (301)496-9275

October 17, 1977

The Honorable Harrison A. Williams
Chairman
Committee on Human Resources
United States Senate
Washington, D.C. 20510

Dear Mr. Chairman:

The Commission for the Control of Huntington's Disease and Its Consequences is pleased to submit its report in compliance with Public Law 94-63. Volume I of the report describes the findings and recommendations which make up the Commission's National Plan for the Control of Huntington's Disease. Companion volumes provide more technical data and documentation.

During the course of its deliberations, the Commission worked closely with scientists, public officials, health care providers, and Huntington's disease patients and families. The plan which evolved addresses the very real needs of these families and many others faced with long-term genetic or neurologic illness. The Commission has sought practical ways to serve the interests and needs of more than one group of patients whenever possible so that the plan can benefit many millions of Americans.

The Commission is grateful for the opportunity to prepare this report. We acknowledge the advice and support of concerned citizens within and without the government and express the hope that the recommendations presented will be the impetus for improvements in the nation's health care system.

Respectfully,

Marjorie M. Guthrie

Marjorie M. Guthrie
Chairwoman

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COMMISSION FOR THE CONTROL OF HUNTINGTON'S DISEASE AND ITS CONSEQUENCES
Building 31, Room 8A11 National Institutes of Health Bethesda, Maryland 20014 (301)496-9275

October 17, 1977

The Honorable Harley O. Staggers
Chairman
Committee on Interstate and Foreign Commerce
House of Representatives
Washington, D.C. 20515

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Building 31, Room 8A11 National Institutes of Health Bethesda, Maryland 20014 (301)496-9275

October 17, 1977

The President
The White House
Washington, D.C. 20500

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Marjorie M. Guthrie

Marjorie M. Guthrie
Chairwoman

Acknowledgements

In the course of its deliberations the Commission was entrusted to speak for the lives and hopes of thousands of individuals across the country who must struggle with Huntington's disease. It has been a privilege and a responsibility to represent these patients and families. It is difficult to find words adequate to express our gratitude to them, and to the many individuals, within and without the government, who gave so generously of their time and talents. We cannot mention all by name, but these persons in particular earn our special thanks:

Dr. Donald B. Tower, Director of the National Institute of Neurological and Communicative Disorders and Stroke, and Dr. Thomas Chase, Director, Intramural Research in the National Institute of Neurological and Communicative Disorders and Stroke, who provided generous and unflagging support as well as wisdom and experience, and their colleagues, Dr. Katherine Bick, Assistant to the Director, Neurological Disorders Program, Dr. J. Kiffin Penry, Director, Neurological Disorders Program, and Dr. George Murray, Director, Office of Program Planning and Evaluation, and other members of his staff for their guidance, advice, and encouragement.

Dr. Ruth Kirschstein, Director of the National Institute of General Medical Sciences, and Dr. Fred Bergmann, Director, Genetics Program, in the National Institute of General Medical Sciences, for their assistance not only to this Commission but also for their dedication to helping all those afflicted with hereditary disorders.

Dr. Bertram Brown and colleagues in the National Institute of Mental Health; Mr. Dennis Tolsma and colleagues of the Center for Disease Control; and Dr. Warren Huber of the Veterans Administration.

Special thanks are also due to:

Dr. Julius Richmond, Assistant Secretary for Health, Department of Health, Education, and Welfare, for his time and attention at the beginning of an exceedingly busy term.

The Honorable Patricia Harris, Secretary of Housing and Urban Development, and Mr. David Williamson, Director, Office of Independent Living for the Disabled at the Department of Housing and Urban Development, for their excellent advice on programs for the handicapped.

The staff of the Pan American Health Organization for facilitating the Commission's international efforts.

Many members of Congress have given unstinting support in the fight against Huntington's disease. Among them we wish to thank especially Senators Birch Bayh, Dick Clark, Alan Cranston, Edward M. Kennedy, Warren Magnuson, and Harrison Williams, and Representatives Tim Lee Carter, Paul Rogers, and Robert Roe.

Staff Assistants Dr. David Blumenthal, Ms. Louise Bracknell, Mr. Jay Cutler, Ms. Barbara Dixon, Mr. Scott Ginsberg, Ms. Marian Laster, Ms. Louise Ringwalt, and in particular, Mr. Terry Lierman, were especially helpful.

A special debt is owed to Mr. Nik Edes, Deputy Undersecretary of Labor for Legislation and Inter-governmental Relations, for advice, concern, and encouragement; and to Dr. Gilbert Omenn, Assistant Director for Human Resources and Social and Economic Service, Office of Science and Technology Policy, for his support and expert counsel. Thanks also to Ms. Sara Seanor for facilitating the work of the Commission.

The Commission staff deserves thanks for heroic efforts beyond any requirement of duty. Ms. Elizabeth McDonald, Administrative Assistant, and Ms. Bette Walling worked under intense pressure with grace and humor. Mr. Joseph Barnard, Administrative Officer, greatly facilitated the work of the Commission.

Others who deserve special thanks are:

Ms. Joan Wilentz, whose talent and sensitivity as a writer are matched only by her willingness to strive for excellence.

Mr. James Booth, Ms. Pauline Gingras, Ms. Sharon Weiner, Ms. Catherine Spanbauer, and other staff of Capital Systems Group, Inc., for their long hours in preparing meetings and in assembling the report.

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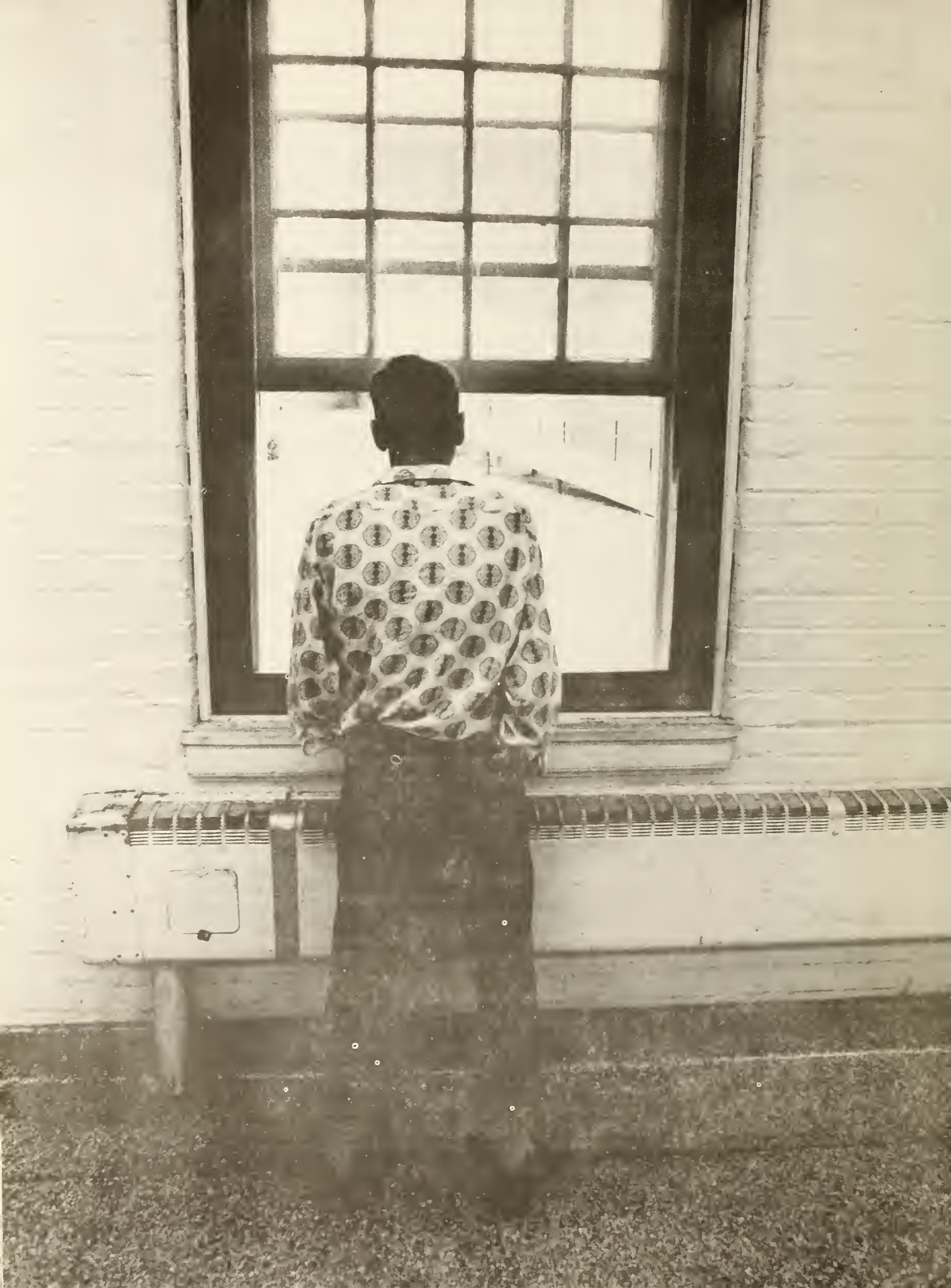
Dr. Judith and Mr. James Walters for their generous assistance.
Ms. Maya Pines for invaluable editorial advice.
Mr. Eugene Kone for his professional expertise and friendship.
Ms. Elizabeth Benson for her gracious hospitality and moral support.

The Commission also extends its gratitude to the scores of government officials, scientists, health care professionals and others who advised and encouraged. Most particularly the Commission is indebted to the Huntington's disease health voluntary organizations: The Committee to Combat Huntington's Disease, The Huntington's Chorea Foundation, The Hereditary Disease Foundation, and The National Huntington's Disease Association, and to all the patients and families affected by Huntington's disease who testified in public hearings and who moved, touched, and spurred the Commission in its work.

Finally, the Commission expresses its deep appreciation to Deputy Director Dr. Charles MacKay for his consummate skill in synthesizing mountains of data, and his contributions to the Commission's work. To Executive Director, Dr. Nancy Wexler, go very special and warmest thanks. Her deep personal commitment and her intelligence, graced with wit, are a force which can make all things possible.



Prologue

The Commission for the Control of Huntington's Disease and Its Consequences was mandated by Congress to study the state of the art of medical and social management in Huntington's disease and to develop a comprehensive national plan for its control. The past year has been devoted to detailed and intense study.

The Commission has concluded that Huntington's disease cannot be studied in isolation. Nor can the solutions, if they are to be truly effective, concern Huntington's disease alone. A disease which is hereditary, which produces radical physical changes—destroying coordination, speech, and self-control, as well as profound mental changes—demolishing the power to think or remember or reason—which causes severe mental illness, strikes in the prime of life, and is progressively fatal over 10 or 20 years—is a microcosm of the major health problems the country is facing today.

The Commission had a very specific charge to study Huntington's disease. It has fulfilled that charge. But in so doing, the Commission found it necessary to expand the scope of its deliberations to consider the more general problems created by hereditary, neurologic, psychiatric, chronic, and terminal disease.

There are, of course, features of Huntington's disease which make it unique. The constellation of mental and physical symptoms, the insidious onset of the disorder, the torment of those at risk waiting throughout their lives to learn if they have been spared, the necessity to make major life decisions in an atmosphere of uncertainty and ambiguity—all conspire to make Huntington's disease "one of the most diabolical diseases known to man," as it is described in many medical texts. There is almost no aspect of life which Huntington's disease or its threat does not touch. There are no relatives of a Huntington's disease family, close or distant, whether genetically involved or not, who can remain unaffected. As a deadly and inherited disorder, Huntington's disease has a cataclysmic and shattering impact on families.

There is no yardstick to measure human misery. Huntington's disease will not be cured because people suffer more greatly, but because scientists supported by generous funding will struggle to find the answers. Huntington's disease patients will find appropriate care in the community not just because they are needy, but because they stand with other groups—the elderly, those with epilepsy, multiple sclerosis, muscular dystrophy, and others—in now demanding a change in long-term care.

The Commission believes that in an era of scarce resources and increasing demands, costly and redundant programs for individual diseases are no longer feasible. Accordingly, the National Plan developed by the Commission adopts a generic rather than a categorical approach, one that is problem-oriented rather than disease-oriented. Recommendations for the control of Huntington's disease and its consequences emphasize the commonalities that unite interests rather than the differences which separate them.

In a very few instances, the recommendations apply to Huntington's disease alone. Other recommendations call for pilot programs designed for Huntington's disease and related disorders, such as Parkinson's disease, amyotrophic lateral sclerosis, stroke, dystonia, multiple sclerosis, presenile and senile dementia, ataxia, and other hereditary and neurological conditions. These pilot programs are to be evaluated after a three-to-five-year period to determine if the recommendations are, in fact, as helpful and practical as they were intended. If so they can be adopted on a larger scale. Some recommen-

dations require major changes that will affect a broad range of persons with a wide variety of disabilities. Most recommendations can be accomplished using existing authorizations and programs. Some require an expansion or readjustment of existing programs. A few demand new legislation and major program development.

The Commission was an interdisciplinary group chosen because of personal involvement or professional interest in the pursuit of a common goal. After a year's study, it was clear that all those concerned with the amelioration of suffering caused by hereditary, neurological, and neuropsychiatric disorders must find common ground. The territorial behavior that sets one disease sufferer against another must give way to a united approach aimed at achieving compassionate care, treatment, and cure for all these diseases.

Congress has granted the Commission a superb opportunity to review the problems created by one complex and formidable disease. This review has entailed a careful look at the state of the art not only in Huntington's disease but in other genetic and chronic disorders. *The Commission has concluded that to generate greater creativity and productivity in biomedical research, provide a more rational and humane plan of health care management, demonstrate greater fiscal responsibility and cost effectiveness, and recognize the needs of countless sufferers from kindred disorders who have no such privileged voice, its recommendations must go beyond Huntington's disease to address a broader range of fundamental health problems.*

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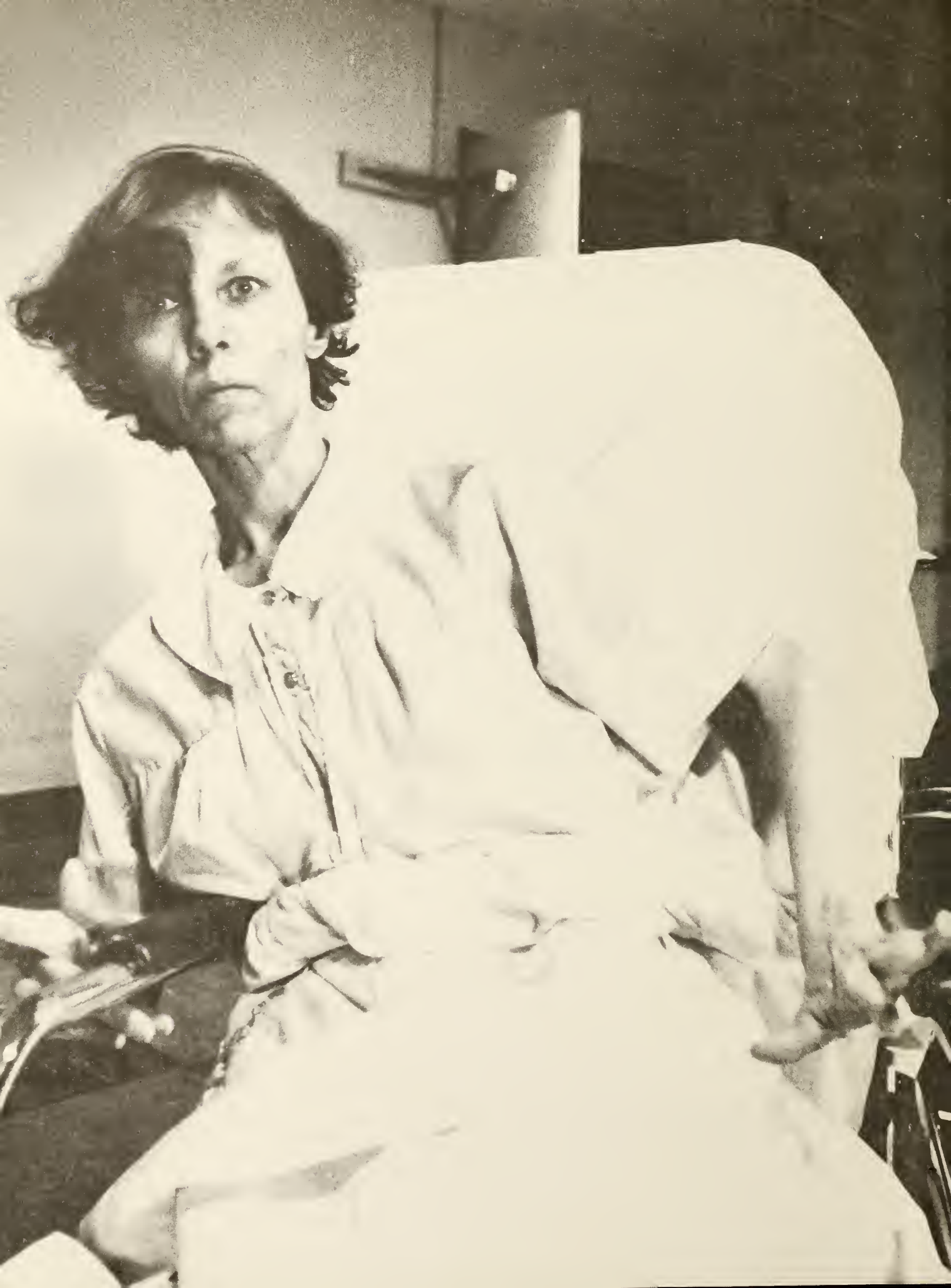


Summary

Huntington's Disease

The fear of losing one's mind and the fear of losing control over one's body are among the most profound fears known to mankind. Both losses occur in Huntington's disease, a hereditary and terminal brain disorder which begins insidiously, usually in middle age. Men and women affected grow irritable or hostile; their personalities change. Some become manic, some apathetic, some suicidally depressed. Powers of reason, memory, and judgment fade, leading inevitably to dementia.

Physical symptoms accompany the mental changes. Patients may appear clumsy or fidgety at first. They may grimace or smile in a peculiar way. As the disease progresses they may have difficulty in talking or swallowing; they lose control over normal body functions. Their restless movements become exaggerated and incessant. They twist, writhe and lurch; they make flailing movements of head, trunk or limbs, ever on the verge of falling.

These symptoms describe what Huntington's disease looks like in someone who has struggled 10 or 20 years with it. It may explain why a woman now known to have had Huntington's disease was hanged as a witch in 17th century Salem. Or why the late composer and folk singer, Woody Guthrie, was considered an alcoholic and was in and out of mental hospitals for years before the true diagnosis of Huntington's disease was made.

Drawing by P. Breughel the younger (1569 A.D.) entitled "These are the pilgrims who have to danse on St. Johnsday in Meulebeeck outside Brussels and if they have danced or jumped over a bridge they are cured for a full year from St. Johns illness" (no. 16859, Rijksmuseum, Amsterdam). Reproduced through the courtesy of the Rijksmuseum, Amsterdam and L. N. Went, M.D., The Netherlands.

More About the Disease and Its Consequences

Inheritance—Each child of a parent with Huntington's disease has a 50-50 chance of inheriting the disorder. There is no way of knowing who has inherited the gene for Huntington's until the symptoms appear.

Age of Onset—The usual time of onset is between 35 and 45, but about 10 percent of cases occur in young people under 20. Children as young as two and adults as old as 80 have been known to develop the disease. People who are "at risk"—who have a parent with the disease—may have to wait a lifetime to know if they have been spared.

Diagnosis—Huntington's disease is usually diagnosed on the basis of the characteristic uncontrollable movements, called *chorea* (from the Greek word for dance), and a family history. Mood or personality changes and minor movements may precede diagnosis by 10 years or more. Some patients show more severe mental symptoms; others have more severe movement problems.

Prognosis—Huntington's disease is chronic, progressive, and terminal, taking its toll over a 10 to 20 year period. There are no states of remission as in multiple sclerosis or some cases of cancer. The disease is usually more severe in children. There is some evidence that the later the disease appears the milder is its course.

Death—Cause of death in Huntington's patients is commonly heart failure, pneumonia, or other infections following an exhausting siege of illness. Accidental death from falls or death from choking, because of difficulties in chewing or swallowing, is also frequent.

Treatment—There is no effective treatment for Huntington's disease. Some medications used in treating schizophrenia provide some relief for the abnormal movements. Other drugs may relieve some of the symptoms of depression or other mood disturbances. No drug can treat the loss of mental faculties. All drugs provide only partial relief in some people for limited times only.

Suicide—There is a high suicide rate among Huntington's disease patients and those at risk. Some authorities estimate it is 7 times the national suicide rate—1.3 percent of all deaths in the United States.

Prevalence—No reliable figures are available on the incidence or prevalence of Huntington's disease. Patients often are misdiagnosed as schizophrenic, or as suffering from Parkinson's disease or other neurologic disorders. Death certificates frequently do not record Huntington's disease as a cause of death. Some families also hide the existence of the disease out of shame, fear, or guilt. Current estimates put prevalence rates in the range of 4 to 7 per 100,000, although some recent studies suggest it may be as high as 10 in 100,000. Approximately twice this number are at risk for the disorder.

Financial Cost—Families affected by Huntington's disease lose income if the patient is the breadwinner, or must pay for domestic help and child care if the homemaker falls ill. They must pay for home care or institutional costs for upwards of 20 years. At the current price of $12,700 a year for nursing home care, costs can mount to $200,000 over 15 years. The costs of long-term care for Huntington's disease patients are so high that *health agency personnel* sometimes advise the healthy spouse to get a divorce so that the patient can become a ward of the state and become eligible for medical benefits. Tragically, that may be the only way the well spouse can attempt to save to meet the future needs of children at risk.

Huntington's disease is a family disease. Every member of the family is affected—emotionally, physically, socially—whether patient, at risk, or spouse. And the disease occurs not once, but over and over again in successive generations.

The National Plan: Philosophy

Huntington's disease is a unique and seriously neglected disorder which merits attention in its own right. So extraordinary is the range of symptoms, however, and so complex are the service needs of patients and families afflicted, that Huntington's disease can serve as an excellent prototype for major neurologic, genetic, and psychiatric disorders which affect millions of Americans.

Neurological and Movement Disorders—More than four million people in the U.S. suffer from serious limitation of activity as a result of neurological disorders. Neurological and visual impairments were the third leading cause of deaths and the leading cause of limitation of activity in the U.S. in 1971. Huntington's disease is a neurological illness which is frequently misdiagnosed as Parkinsonism, ataxia, and less well-known movement disorders such as torsion dystonia, tardive dyskinesia, and hereditary tremor. Patients with Huntington's disease share symptoms with those suffering from degenerative disorders such as multiple sclerosis, the muscular dystrophies, and amyotrophic lateral sclerosis (Lou Gehrig's disease). Research findings or techniques developed in the study of one disease can illuminate other disease processes. The potential for generating useful information is greatly expanded if patients with Huntington's disease and related disorders serve as controls for each other in experiments.

Genetic Disorders—An estimated 6 percent of the population—over 12 million people—suffer from severe genetic disease. The figure increases dramatically if diseases which have some hereditary component—such as schizophrenia, diabetes, high blood pressure, coronary heart disease, and certain cancers—are included.

Of the 2,300 known genetic disorders, 500 are neurological. Huntington's disease also belongs to the group of nearly 1,000 "dominant" gene disorders (only one abnormal gene is needed to produce illness). Examples of dominant disorders are achondroplasia (dwarfism), cerebellar ataxia, familial hypercholesterolemia (a disorder that predisposes to early heart disease), and polyposis (a condition that leads to cancer of the colon). If the underlying defect in Huntington's disease could be discovered, it might shed light on the genetic abnormality operating in other dominant disorders.

Psychiatric Illnesses: Schizophrenia, Manic-Depressive Psychoses and Personality Disturbance—The mood swings and personality changes that occur in Huntington's disease occur in the major psychoses found throughout the world. Huntington's disease patients exhibit the impatience, limited attention span, and lack of judgment found in the severely mentally ill. They hallucinate and experience delusions, paranoid feelings, and suicidal depressions. Many experience moderate but chronic depressions. Explaining the biological mechanism underlying Huntington's disease might expand understanding of the major psychiatric cripplers and lead to new and better medications. It might also elucidate the biochemistry of moderate depression—a phenomenon of virtually epidemic proportions in this country.

The Diseases of Aging: The Senile and Presenile Dementias—Huntington's disease is classified as a "presenile dementia." This means that Huntington's disease patients experience the same decline in mental faculties that occurs in senility—only they become senile before they are old. Some Huntington's disease patients also seem

to age rapidly, looking older and more feeble than their years. An understanding of the loss of intellect in Huntington's disease might provide insight into some of the diseases of aging—with the potential that the most dreaded aspect of aging, senility, would be ameliorated.

The Commission has concluded that the program for research recommended in the National Plan for Huntington's disease must be basic and broad. Understanding Huntington's disease—with the goal of treatment, prevention, and, ultimately, cure—will entail fundamental insights in the workings of the human nervous system and the mechanisms of inheritance. Fruitful leads may come from experiments directed at Huntington's disease alone (targeted research). But medical history shows that often the answer to one disease comes from studies of other related disorders or even from seemingly unrelated areas.

Different Diseases—Common Service Needs

Research is not the only area uniting Huntington's disease with other disorders. Patients with chronic, progressive, hereditary, and neurological diseases have similar problems in care and treatment. They have common financial, legal, ethical, and social concerns. Huntington's disease patients and families are part of a larger community of Americans frustrated, confused, and in despair over inadequacies and inequities in the country's health care system.

All families faced with serious or terminal illness—whether due to Huntington's disease, multiple sclerosis, stroke, or other diseases—need a *coordinated* system of health care services to provide a *continuum of long-term care* from household aid to institutional care. They need *financial relief* from the bankrupting costs of chronic care which reduce middle income families to poverty. Patients and families alike need *support and guidance from skilled mental health workers.* Those with hereditary illnesses need *complete genetic information given in a supportive environment.* They need *health care providers and health agency personnel who understand the nature of chronic genetic or neurological illness.* The health care system must not abandon patients it cannot cure. Symptoms can be mitigated or disease progression forestalled by programs that emphasize compassionate caring where curing is not yet possible.

This means recognizing the rights of all individuals with chronic and progressive illnesses, and extending to them financial aid and services which have been denied because of restrictive eligibility requirements or arbitrary interpretations of the law. It means coordinating and publicizing services so consumers and personnel are aware of what is available. And it means generating new services or facilities where the demand is great but the supply non-existent.

A Generic Approach

The common research and service needs of Huntington's disease patients and others with chronic hereditary or neurological illness underlie the Commission's decision to adopt a generic, problem-oriented approach in its National Plan rather than one which is categorical—based on the needs of a single disease. The health care system in the United States cannot afford to treat every disease independently. Manpower, equipment, and facilities are limited. By pooling resources, research, care, and treatment programs can be developed to apply to many disorders—no matter how small or scattered the individual populations of patients might be.

The National Plan: Major Findings and Recommendations

RESEARCH

The urgent and overwhelming plea from Huntington's disease patients and families across the nation was for increased support of biomedical research. This critical recommendation dominated all others.

At a time when the neurosciences and genetics are acknowledged to be at the leading edge of science (almost half the Nobel prizes in physiology and medicine in the last 16 years went to neuroscientists) there has been a serious decline in funds for the very institutes of the National Institutes of Health which sponsor such research. The National Institute of Neurological and Communicative Disorders and Stroke, the leading agency for research in the neurosciences, could fund less than one-fourth of approved grants in 1977—only three of these grants concerned Huntington's disease specifically. The Institute's share of the total National Institutes of Health budget was only 6.5 percent in 1977. The National Institute of Neurological and Communicative Disorders and Stroke appropriations do not even keep pace with inflation.

The National Institute of General Medical Sciences supports fundamental research and research training in genetics and in molecular and cell biology. Since 1968, 10 Institute-supported scientists have won Nobel prizes. The Institute has sponsored research which has revolutionized understanding of the basic processes of life in heredity and the behavior of the living cell. Yet in 1977 the Institute could fund only about one-fourth of approved grants; it received only 10 percent of National Institutes of Health funds. Estimates for 1978 project some improvement in funding levels, but major and continuing expansion in the field is also anticipated.

All the scientists who counseled the Commission, including Nobel laureates and world leaders in the neurosciences and genetics, emphasized the need for increased support of a *broad program of basic and clinical research.*

The brains of patients who have died with Huntington's disease show characteristic degenerative changes and nerve cell destruction in areas concerned with thinking, memory, emotion, and the control of movement. Associated with these losses are diminished amounts of essential "neurotransmitters"—chemicals vital to the conduction of the nerve impulse.

Research is needed to determine if the changes in neurotransmitter chemistry are the primary defect caused by the gene for Huntington's disease. It is also possible that some other gene-controlled activity may be involved, such as alterations in cell membranes which affect a cell's ability to function properly. These cell changes might occur in organs of the body outside the brain. The disciplines of the neurosciences and of genetics are critical in this basic research.

The research also has practical ramifications in the search for safe and effective drugs which might replace the chemicals found to be deficient in Huntington's disease patients. It is also vital in the quest for a safe and reliable test (using blood or other easily accessible tissue or fluid) which would distinguish carriers from non-carriers of the Huntington's disease gene before symptoms appear. *A presymptomatic detection test would be a major step in preventing and eventually eliminating Huntington's disease.*

Recommendation: Congress must increase appropriations to support basic and clinical research on the nervous system, in genetics, and in other areas relevant to an understanding of Huntington's disease and other chronic genetic, neurologic, and psychiatric illnesses and the diseases of aging. These appropriations should support research at appropriate institutes of the National Institutes of Health such as the National Institute of Neurological and Communicative Disorders and Stroke, the National Institute of General Medical Sciences, and the National Institute on Aging, as well as other bureaus, institutes, and divisions of the Federal government such as the National Institute of Mental Health and the Veterans Administration.

SPECIAL RESEARCH ACTIVITIES

The following section focuses on special populations, resources, and research aids of outstanding value in the study of Huntington's disease and related disorders.

The Venezuela Project—There is a unique concentration of Huntington's disease patients, descended from a common ancestor, living in rural communities near Lake Maracaibo in Venezuela. Investigators believe that the study of these special individuals would provide a rare opportunity to discover the basic biological mechanism of Huntington's disease. Intermarriage within the group may have produced individuals who have inherited the defective gene from both parents. The effect of two abnormal genes might be more apparent in chemical or other tests.

Recommendation: Congress should appropriate funds to the National Institute of Neurological and Communicative Disorders and Stroke to enable that agency to collaborate with Venezuelan scientists to design, support, and conduct and interdisciplinary study of the population affected by Huntington's disease in the state of Zulia, Venezuela.

National Tissue Bank—Research on the human nervous system requires a source of carefully collected, stored, and documented tissues and biological fluids from patients who have died with nervous system disease. Such tissue, as well as normal tissue, is essential in the study of Huntington's disease and other neurological and psychiatric disorders for which there is no animal model. The tissues of choice are brain, which, usually, can only be collected at autopsy, and cerebral spinal fluid. These tissues are not available in the quantities needed by investigators, nor are methods of collection, preservation, and storage standardized.

Recommendation: Congress should provide funds and direct the Secretary of the Department of Health, Education, and Welfare to establish a National Tissue Bank for the collection, preservation, and distribution of tissues for research on Huntington's disease and related neurological and psychiatric disorders.

Voluntary Patient Rosters—Carefully maintained, accurate, and up-to-date records of patients who have volunteered to participate in research projects are vital to the conduct of clinical research.

Recommendation: Support should be provided by contract with the National Institute of Neurological and Communicative Disorders and Stroke for the maintenance of lists of patients and their families who consent to be listed. These rosters could be the responsibility of state or private health agencies (health voluntary organizations or medical schools). Provision should be made to protect the privacy and confidentiality of those listed.

Interdisciplinary Workshops—Ways to generate new ideas, exchange views across disciplines, and communicate findings quickly are especially important in rapidly developing fields such as genetics and the neurosciences. Interdisciplinary workshops are an effective and economic way of bringing challenging problems into sharp focus and catalyzing activity to solve them. They encourage imaginative speculation, rapid information exchange, and can be instrumental in stimulating new career orientations for young scientists. Interdisciplinary workshops in Huntington's disease have already produced new research hypotheses, demonstrated new technologies, and attracted young as well as well-established scientists to the field of neurogenetic disease research.

Recommendation: The National Institute of Neurological and Communicative Disorders and Stroke should provide financial support, through its grant or contract mechanisms, for interdisciplinary workshops conducted by a Government or voluntary agency, to generate and explore promising leads in research, care, and treatment of Huntington's disease and related disorders.

Neurogenetic Newsletter—Existing journals do not satisfy the need for rapid communication among research scientists and clinicians interested in Huntington's disease and related disorders. An informal publication which would report brief remarks, negative findings, and news of current research and health care projects would be a practical means of circulating information in a growing field.

Recommendation: The National Institute of Neurological and Communicative Disorders and Stroke should sponsor, through its grant or contract mechanisms, a publication for professionals designed to provide brief bulletins on research, management, and related information for individuals interested in Huntington's disease and other neurogenetic disorders.

Conferences and Symposia—A formal review of the state of the art in Huntington's disease research, treatment, and patient care is needed periodically to consolidate the field.

Recommendation: The National Institute of Neurological and Communicative Disorders and Stroke should sponsor an international symposium on Huntington's disease within the next fiscal year (FY 1979). Additional symposia should be held every five years as long as such forums prove useful.

Small Grants Program—"Seed money" to test a promising hypothesis, experiment with new technologies, or learn new techniques is often difficult to obtain under current funding practices of the National Institutes of Health. Yet pilot data are usually necessary to support a grant application. Small grants which can be expedited to an investigator will permit research for a limited period of time, to determine the feasibility of full-scale funding of a project.

Recommendation: Congress should establish a small grants program in the National Institute of Neurological and Communicative Disorders and Stroke to foster pilot or small-scale projects to explore new scientific technologies or test the feasibility of innovative research hypotheses in the neurosciences in general.

General Clinical Research Centers—The General Clinical Research Centers program maintains special units, usually in medical teaching institutions, where clinical research can be conducted. The centers, averaging 10 patient beds each, provide a unique resource for investigators to study many different disease problems using common resources. Research conducted through center programs has led to new insights into disease processes and speeded the application of new research findings to patient treatment and care.

Funding for the General Clinical Research Centers programs has been held at a constant level of $42 million since 1972. As a result, the number of patient beds has declined from 907 in 1972 to 779 in 1976. This decline threatens the future of clinical research. Funding must be increased to maintain high standards and ensure that young physicians will be attracted to clinical research as an alternative to clinical practice.

Only $50,000 of General Clinical Research Centers funds was spent on Huntington's disease research in 1976. This amount must increase as more investigators become active and need to study patients with Huntington's disease and related disorders.

Recommendation: Funding for the General Clinical Research Centers program should be commensurate with clinical research needs and should take into account not only the rising costs of inflation but also the value this program offers as a service delivery mechanism.

Research Training—Training programs for talented investigators with innovative ideas are necessary to produce sustained and high-quality research. There has been an ominous drop in support for research training programs, from a high of $169 million in FY 1969 to $122 million in FY 1976. The number of persons in training programs is down 25 percent, from a high of 14,122 in 1969 to 10,546 in 1976.

Diminished research support and certain "pay-back" provisions of the National Research Service Awards Program have had a harsh impact on clinical research. In 1971 there were 4,779 post-M.D. degree holders receiving National Institutes of Health research training stipends and fellowships in clinical research. Four years later, in 1975, the number had dropped to 2,797—a 40 percent decline. Acute shortages of clinical researchers in the future mean decreases in research directly aimed at developing new treatments for diseases.

Recommendation: To ensure a continuous supply of highly qualified and well-trained investigators it is essential that:

- Annual levels of appropriations to the National Institute of Neurological and Communicative Disorders and Stroke for research training under the National Research Service Awards Program be set at $9.3 million in FY 1979, $11.7 million in FY 1980, and $12.9 million in FY 1981.

- Annual appropriations for the National Institute of General Medical Sciences for research training under National Research Service Awards (including Minority Access to Research Careers) be set at $46 million in FY 1979, $48.3 million in FY 1980, and $50.7 million in FY 1981.

- Annual appropriations for research training to the other institutes conducting research related to Huntington's disease (National Institute of Mental Health and National Institute on Aging) be increased.

DRUG DEVELOPMENT

The development of drugs to treat uncommon diseases is effectively blocked because small markets fail to excite the commercial interest of the pharmaceutical industry.

Many hereditary and neurological diseases affect relatively small populations but in the aggregate, they are responsible for a major portion of the mortality and morbidity of this nation.

Drugs, especially central nervous system drugs which are developed to treat neurological disorders such as Huntington's disease, may have much wider applications—as sedatives, tranquilizers, and antidepressants.

Recommendation: The President should immediately appoint an interdepartmental Task Force to consider ways of encouraging and accelerating the development of new drugs to meet the urgent needs of small populations of patients afflicted by crippling or lethal diseases. The Task Force should include representatives of the Departments of Commerce, the Treasury, and Health, Education, and Welfare, as well as members of Congress, representatives from the pharmaceutical industry, and consumers.

CENTERS WITHOUT WALLS

There are *no* clinical facilities specifically devoted to research, care, and treatment of patients with Huntington's disease or related disorders.

There *are* individual investigators in universities or medical centers throughout the country conducting basic and clinical research on these diseases.

There is *no* need to constrict researchers and patients to one building or to a confined geographical area—as traditional centers are constructed.

There *is* a need to link scientists with a clinical facility, and there *is* a need for patients with Huntington's disease or related disorders to have access to a clinical facility offering integrated programs of medical, psychological, social, and rehabilitative services. These patients would be able to participate in clinical drug trials and other experimental studies.

There *is* a need to conduct research on the delivery of health care and to demonstrate excellence in care and treatment for patients with Huntington's disease and related disorders, before new treatments and care programs are disseminated into the community.

Recommendation: Congress should appropriate funds and direct the Secretary of Health, Education, and Welfare to establish two demonstration projects of "Centers Without Walls." Each center should include a clinical facility for conducting basic and clinical research on patients with Huntington's disease and related disorders, as well as research on services (speech and physical therapy, mental health, genetic counseling, etc.). The clinical facility, which would serve as the administrative core, would be affiliated with center investigators engaged in basic and clinical research elsewhere in the country. The Center should function as a demonstration, education, and training model in research, treatment, and care of Huntington's disease and related disorders. Specific demonstration authority for the National Institute of Neurological and Communicative Disorders and Stroke would facilitate conducting certain studies.

HEALTH SERVICES COORDINATION AND DEVELOPMENT

Health services and programs which could benefit Huntington's disease patients and families, and others with chronic disabling diseases, are so fragmented that many who are eligible receive no help. Others with similar needs are considered ineligible. The problem pervades the health care system at the local, state, and Federal levels.

A number of existing community services, such as vocational rehabilitation and Community Mental Health Centers, could be adapted to meet the needs of Huntington's disease patients and families. Other services may not exist but could be developed for a group of disorders, such as group homes, respite care, and Work and Recreation Day Centers.

Recommendation: Congress should appropriate funds and establish positions for two pilot programs of community Health Services Coordination and Development. These should be initiated as a collaborative project between the Center for Disease Con-

trol and the National Institute of Neurological and Communicative Disorders and Stroke. The purpose of the programs should be to integrate services, ensure access to services by providing patient advocates or ombudsmen, and develop new services where none exist. This project should be closely integrated with Centers Without Walls.

FINANCIAL DEVASTATION

Long-term care is the major cause of catastrophic health expenses in the United States today.

The middle income family affected by chronic illness has little or no financial protection and is quickly impoverished. The spouse of a Huntington's disease patient is often advised to get a divorce in order to maintain the home and save for the future needs of at-risk children. The ill spouse then becomes indigent—and eligible for medical benefits.

The direct costs for Huntington's disease are approximately $62,500 to $250,000 *per patient* over a course of 20 years. According to conservative National Institute of Neurological and Communicative Disorders and Stroke estimates, Huntington's disease costs about $80 million a year. Other Commission estimates range from $110 million to $125 million annually for direct costs. This does *not* include productivity lost, the cost of extra housekeeping, special food, clothing, safety devices, aids to the handicapped, rehabilitation, and other services. This also does *not* include the price of social and emotional upheaval which can lead to additional medical and psychotherapy costs for family members.

Private and government financial aid programs are not designed for the chronically ill.

Certain Social Security programs designed to aid families in financial need have the effect of discouraging work and splintering families.

Private medical insurance rarely pays long-term costs. Some insurance companies explicitly exclude coverage for Huntington's disease. Most will not pay for "non-treatable" illness or custodial care.

Medicare is geared to the acute hospital system and does not pay for chronic care.

Medicaid provides protection against catastrophic expenses, but only for the indigent or near indigent. Benefits vary widely from state to state. The designation "medically needy" provides some measure of relief for persons whose income is too high to qualify for Medicaid, but too low to cover medical expenses. Individuals must still "spend down" to near the poverty line to qualify, and coverage is minimal and highly erratic. Although Medicaid is theoretically a comprehensive service program, in practice it is oriented to skilled services in an institutional setting. Over 98 percent of all Medicaid long-term care expenditures are for nursing home care—Medicaid finances 77 percent of all government-sponsored long-term care.

Supplemental Security Income provides some monthly income and qualifies an individual for Medicaid benefits. Again, families must have incomes at or near the poverty level to qualify. If the healthy spouse earns more than the specified income

level, Supplemental Security Income is lost, and with it, eligibility for Medicaid. Medical costs may be so exorbitant that the family is automatically reduced to the poverty level in attempting to pay. A family receiving Supplemental Security Income cannot save for any health or education needs for other family members, including children who are at risk.

Disability Insurance under Title II of the Social Security Act provides some financial assistance for wage earners. To qualify, certain work criteria must be satisfied. Because the onset of Huntington's disease is insidious and the first signs are frequently psychological, many patients have an irregular work history and do not qualify. *Women with chronic disease who are homemakers are ineligible because their labor in the home has not been for wages.*

A disproportionately lower number of Huntington's disease patients receive benefits under Disability Insurance and Supplemental Security Income than would be expected in comparison with other neurologic disorders, based on estimated prevalence rates. It is not known if patients are mistakenly disallowed, or if they are never informed of the program. The same state agencies using the same medical criteria make the determination for both programs.

Title XX of the Social Security Act provides funds to states for developing social services, including those needed by the disabled such as homemaker and chore assistance, and day care. Yet services are limited and only low income groups are eligible. Few disabled persons receive help.

Recommendation: The President, the Congress, and the Secretary of Health, Education, and Welfare should ensure that the needs of the chronically ill are addressed in any program of National Health Insurance. Coverage of long-term care in such an insurance program should include the full range of medical, social, and mental health services required by the chronically ill.

The Secretary of Health, Education, and Welfare, through the Commissioner of the Social Security Administration, should immediately take the following steps to correct deficiencies in the operation of programs authorized under the Social Security Act as amended:

New medical guidelines for determining disability under Title II (Disability Insurance) and Title XVI (Supplemental Security Income) should be issued, incorporating the most recent findings about the nature and course of Huntington's disease. These guidelines should be distributed to Social Security Administration regional and district offices and to state vocational rehabilitation offices.

A program of centralized review of claims for allowances under Title II and Title XVI, on the basis of disability due to Huntington's disease, should be initiated to assure quality control and uniformity of decisions.

Regulations regarding Title XVI should be modified to exclude, in determining resources of individuals with permanent disability leading to death, moderate savings placed in trust for specific stipulated purposes, such as education or future medical needs of children, whether placed in trust before or after application for benefits under Title XVI.

Income eligiblity requirements should be revised to permit continued Title XIX (Medicaid) and Title XX (Social Services) benefits to individuals with permanent disability leading to death, until family income exceeds 115 percent of the median family income for the state.

Congress should consider new legislation or amendments to the Social Security Act to address the needs of women who become disabled. Coverage under Title II (Disability Insurance) of the Social Security Act should be extended to persons who have been homemakers and whose labor in the home has not been for wages.

All state agencies administering Social Security programs under Title XIX (Medicaid) of the Social Security Act should extend coverage to those designated as medically needy, and ensure, through vigorous implementation, that all who are eligible receive benefits.

A CONTINUUM OF CARE

Few communities provide a continuum of care—a full range of services and facilities matched to the needs of the chronically ill at all levels of disability. Where suitable care can be found, patients may not have access because of age, high cost, restriction to certain disabling conditions, or ineligibility for a government program.

Because alternatives to institutionalization—home health care, congregate housing, respite care, day centers and the like—are lacking, patients are often prematurely institutionalized. Inappropriate institutionalization can have a demoralizing and destructive effect on the patient as well as impose an unnecessary financial burden.

No existing facilities are appropriate for young and middle-aged persons with Huntington's disease and other movement disorders. Conventional institutions present numerous architectural hazards for patients who fall. Staff is often inexperienced and ill-equipped to handle the special problems of these patients.

Abuses in long-term care facilities are wide-spread and well-documented. They include inadequate training for staff in neurologic and geriatric nursing; deficiency of quality control standards and enforcement mechanisms; and limited attention to the social, emotional, and environmental needs of the patient.

There is a critical need for individuals who can provide transportation, domestic chore, therapist aide, companion, and other services for the homebound disabled.

Recommendation: A continuum of care services and facilities should be available to Huntington's disease patients and others with chronic or disabling conditions, to permit individuals to function at an optimal level commensurate with the degree of impairment. Local Health Systems Agencies and State Health Planning and Development Agencies should ensure, through their Health Systems and Annual Implementation Plans, that health provider organizations develop such services and facilities.

Congress should appropriate funds and designate the National Institute of Neurological and Communicative Disorders and Stroke as lead agency in the implementation

of the following pilot projects, all of which should be coordinated with Centers Without Walls, if possible.

1. *A research and demonstration project for the long-term institutional care of Huntington's disease patients and others who are neurologically impaired.* Innovative programs in long-term care, using staff specially trained in neurological nursing, should be provided. New architectural and interior designs should be explored to determine how best to provide a comfortable and hazard-free environment which is also conducive to keeping neurologically impaired patients mentally alert and oriented.

2. *A respite care program for Huntington's disease patients and other patients living at home.* This program would arrange for regularly scheduled short-term stays for patients in an underutilized community hospital or other health care facility. Patients would receive medical care and have access to other services during their stays, and families would be free of caretaking responsibilities at these times.

3. *Work and Recreation Day Centers for Huntington's disease patients and other chronically ill or disabled persons.* The centers would provide social and recreational activities, rehabilitation programs, a sheltered workshop, and other social or therapeutic services. Staff could include personnel trained under the Comprehensive Employment and Training Act.

4. *New health care personnel pilot programs.* Jobs and services in long-term health care should be developed in several states under the auspices of the Comprehensive Employment and Training Act, utilizing public assistance jobs.

GENETIC COUNSELING

"Traditional" genetic counseling—explaining the nature of the illness, probability of inheritance, and other facts in one or two sessions—is often less useful than it could be. Decisions and attitudes change as patients, individuals at risk, and other family members adjust over a long period of time to living with the disease or the threat of disease.

Little is known about the impact of genetic counseling on individual behavior. Much research is needed to evaluate the effects of genetic counseling in terms of information assimilation, emotional reaction, and influence on such major life decisions as career choice, marriage, and parenthood.

Recommendation: Programs should be supported to train genetic counselors, social and behavioral scientists, and other health professionals sophisticated in genetics and in the requisite counseling skills; genetic counseling services should be developed which provide access to long-term support and follow-up; and research should be conducted on the most beneficial and accessible placement of genetic counseling services, on the nature of the services to be provided, and on the impact of such services. This is to be accomplished under the auspices of P.L. 94-278, Title IV, the "National Genetic Diseases Act."

HEALTH PSYCHOLOGY

Individuals and families confronted with Huntington's disease are at high risk to develop serious emotional problems. All family members experience serious psychological stress in reaction to the condition.

Huntington's disease patients suffer not only from the psychological reaction to the illness but also from emotional disturbances which are part of the disease process. Psychological stress exacerbates mental and physical symptoms.

What is true of Huntington's disease patients and families is true of others faced with life-threatening conditions.

Neurologists usually do not have the time or the training to treat patients' and families' emotional problems, while mental health professionals usually lack training about Huntington's disease—and about organic disorders, in general. As a result, patients and family members have no one to turn to for the knowledgeable support and understanding which could lessen or even prevent psychological complications.

Recommendation: Patients and family members who are affected by Huntington's disease or other chronic neurologic or hereditary disorders should have access to affordable long-term psychological counseling and support. This should be given by trained individuals, knowledgeable about the medical aspects of the disorder and psychotherapeutically skilled, as a standard part of a comprehensive treatment plan to limit or prevent serious emotional disturbance.

Congress should appropriate funds to support pilot and demonstration projects to be initiated immediately by the Director of the National Institute of Mental Health:

A pilot program in *prevention and treatment of mental illness* in patients and family members affected by Huntington's disease and related neurologic and hereditary disorders. This program should be established with the cooperation of five Community Mental Health Centers or with center staff working in liaison with a community-based hospital or medical center.

A demonstration program to review and develop the state of the art in health psychology, beginning with a *Symposium on Psychological Response to Neurologic Conditions: Primary Prevention and Treatment.* This symposium should focus on the psychological problems of chronic neurologic illness using Huntington's disease as a prototype. Ultimately an ongoing workshop program should be maintained to develop training materials, relevant courses, research ideas, and effective communication devices.

EMPLOYMENT

Discrimination in employment against patients with Huntington's disease and individuals at risk has been reported. Many witnesses providing public testimony chose to remain anonymous because of fear that they or their relatives would lose their jobs.

Many Huntington's disease patients and at-risk persons are unaware of Federal affirmative action programs and protections against discrimination in the Rehabilitation Act of 1973, Sections 503 and 504.

As methods for screening for a variety of genetic disorders increase, special precautions are needed to ensure that those who are afflicted with, carriers of, or at risk for a hereditary illness do not become special targets for discrimination. Public education is one such protection.

Productive activity is important for the psychological and physical well-being of patients. Sheltered workshops or Government-industry cooperative projects similar to the Training and Placement Service Program funded by the Comprehensive Employment and Training Act would engage Huntington's disease patients in useful work and would be beneficial to both patient and society.

Recommendation: The Secretary of Labor should ensure that all persons with Huntington's disease and other neurological or otherwise handicapping conditions have the benefit of work suited to their capabilities, from sheltered workshops to full employment.

Guidelines must be developed for physicians who evaluate patients and make job placement recommendations. The support of concerned health voluntary organizations should be solicited in hiring the handicapped.

The Secretary should also guarantee the rights of Huntington's disease patients, those at risk, and others who are either handicapped or perceived to be handicapped, to affirmative action and antidiscrimination protections through stringent enforcement and vigorous outreach as required by Section 503 of the Rehabilitation Act of 1973.

REHABILITATION

Rehabilitation services are oriented to handicapped individuals whose conditions are stable or expected to improve. The primary intent of the program is to return individuals to employment. Patients with progressive and terminal disease—though they may have 10 to 15 years of productive life—are turned away as bad investments.

Recommendation: The Commissioner of the Rehabilitation Services Administration should designate persons with Huntington's disease and related disorders as underserved groups. The initiation and expansion of services to this group should include the training and orientation of rehabilitation counselors regarding Huntington's disease and related disorders. Adequate psychological counseling and assistance in job-finding, or adjusting to living at home or in the community at a maximal level of functioning, should be provided.

EDUCATION

Misdiagnosis of Huntington's disease was one of the most serious problems reported in the public testimony. The need for professional education was repeatedly emphasized by witnesses. Particularly tragic were the stories of those who had had children before an accurate diagnosis of hereditary disease was made. And the problem persists.

Families with Huntington's disease, community service personnel, and the public need to be educated about Huntington's disease to ensure proper care and to prevent stigma and discrimination. A major priority is the development of educational materials to enable patients and families to take an active part in their own care.

Medical and lay education in America in all aspects of genetics is extremely limited. A central source to collect, develop, and disseminate educational materials would facilitate access and would be efficient and cost-effective. Information should concern human genetics, genetic diseases, and birth defects.

Recommendation: Congress should appropriate funds for the development and dissemination of educational materials on Huntington's disease for families, health care professionals, community service personnel, and the public.

A national information clearinghouse should be established for the purpose of developing, collecting, and distributing educational materials on human genetics, genetic disorders, and birth defects, as a means of implementing P.L. 94-278, Title IV, the "National Genetic Diseases Act."

NATIONAL SICKLE CELL ANEMIA, COOLEY'S ANEMIA, TAY-SACHS, AND GENETIC DISEASES ACT, P.L. 94-278, TITLE IV

This Act provides for research and services—screening, counseling, information, and education—for all genetic diseases. While the Act was passed in 1976 with an authorization of $30 million annually for FY 1976-1978, no implementation plan has been developed and no monies have been appropriated. For the first time, the current Labor-HEW appropriations bill includes $4 million for the Act.

Recommendation: P.L. 94-278, Title IV, the National Sickle Cell Anemia, Cooley's Anemia, Tay-Sachs and Genetic Diseases Act, should be extended and the full authorization of $30 million should be appropriated for FY 1979, FY 1980, and FY 1981.

An administrative unit should be established in the Office of the Assistant Secretary for Health which would be responsible for planning, evaluating, and coordinating genetic services and research, monitoring grant review and allocation of funds, establishing program and funding priorities, and planning policy with respect to the social, economic, legal, ethical, and psychological problems that are anticipated with the development of new technologies in genetic screening. Input from non-government professionals and consumers should be solicited. This office should have responsibility for monitoring the implementation of those sections of the Huntington's Disease Commission National Plan which have relevance to the provisions of the Act.

IMPLEMENTATION

All recommendations in the National Plan of the Huntington's Disease Commission can and should be initiated at once. All will require careful follow-up to ensure that the full intent of the National Plan is carried out and that individual provisions are acted on promptly and thoroughly.

Recommendation: A branch for Huntington's disease and related disorders should be created within the Neurological Disorders Program of the National Institute of Neurological and Communicative Disorders and Stroke. This branch would be charged with overseeing the implementation of the National Plan for the Control of Huntington's Disease and Its Consequences. The branch should be represented on the Public Health Service Genetics Coordinating Committee. It should coordinate activities with the proposed Genetic Services and Research Unit in the Office of the Assistant Secretary for Health.

National Plan — Budget Summary

The criteria used in developing the budget were that dollar amounts would be allocated only for programs designed specifically for Huntington's disease patients and families and those with related disorders. Generic programs with widespread benefits are not included, with the exception of the Small Grants Program which is to promote research in the neurosciences in general.

Budget Categories	FY 1979	FY 1980	FY 1981
Research on Huntington's Disease and Related Disorders (Includes Small Grants Program)	$8,900,000	$9,968,000	$11,164,160
Special Research Activities			
Venezuela Project	150,000	143,000	160,160
National Tissue Bank	175,000	150,000	168,000
Patient Rosters	35,000	5,000	5,600
Interdisciplinary Workshops	150,000	168,000	188,160
Neurogenetic Newsletter	30,000	33,600	37,632
International Symposium	80,000	—	—
NIMH Symposium	50,000	—	—
Pilot Programs			
Centers Without Walls — (2)	2,080,000	2,329,600	2,609,152
Health Services Coordination and Development programs (2)	800,000	896,000	1,003,520
NIMH Pilot Program (in 5 CMHCs)	350,000	392,000	439,040
Continuum of Care Programs:			
Respite Care (2)	1,000,000	1,120,000	1,254,400
Model Long-Term Care Facilities (2)	1,200,000	1,344,000	1,505,280
Work and Recreation Centers (3)	300,000	336,000	376,320
Education: Development and Distribution of Materials	400,000	350,000	392,000
Implementation			
Huntington's Disease and Related Disorders Branch	75,000	84,000	94,080
TOTAL:	$15,775,000	$17,319,200	$19,397,504

New Staff Positions: 23 new positions (10 in NINCDS; 13 in CDC).
Total Three-Year Budget Request: $52,491,704.

Overall Budget Increases for NINCDS* and NIGMS*

	FY 1979	FY 1980	FY 1981
NINCDS	**$218,000,000**	**$245,000,000**	**$273,000,000**
Includes Research Training:			
• National Research Service Awards Program	9,300,000	11,700,000	12,900,000
• Research Career Program Awards	3,500,000	4,000,000	4,600,000
NIGMS	**244,000,000**	**259,000,000**	**274,000,000**
Includes Research Training:			
• National Research Service Awards, including Minority Access to Research Careers	46,000,000	48,300,000	57,015,000

*Includes costs of Commission recommendations which are the responsibility of these agencies.

Features of the Plan

In keeping with the generic approach of the Commission, most recommendations apply to *Huntington's disease and related disorders.* Some apply to an even broader group of patients. All pilot programs are to be evaluated after stipulated intervals to determine if they should be continued, altered, expanded, or terminated. Most recommendations can be accomplished using existing authorizations and facilities, some require expansion or readjustment of existing programs, while a few demand new legislation or major program development.

If fully accomplished the Plan will:

- Lead to the detection, treatment, and ultimately, cure of Huntington's disease; and provide compassionate and appropriate care for Huntington's disease patients and their families who are now affected.

- Advance research in understanding how the human brain works, how movement is controlled, and what happens when the essential human faculties of reason, memory, emotion, and judgment are impaired in chronic genetic, neurologic, and psychiatric illness.

- Advance understanding of the mechanisms of inheritance that can lead to the detection and prevention of severe, disabling, and fatal hereditary disease.

- Encourage the development of drugs or other medications to treat a variety of serious and fatal diseases—especially those affecting the central nervous system—in cases where research and development have been hampered because the populations of patients to be served are small.

- Rescue middle income families from the financial devastation wrought by chronic illness.

- Rehabilitate Huntington's disease patients and others with progressive disorders through vocational, physical, and psychological programs to prolong their useful and productive lives and return them to some form of work if they are able.

- Provide genetic counseling to families with Huntington's disease and other genetic disorders so that individuals may exercise free choice regarding major life decisions with full knowledge of the disease and its consequences.

- Provide pilot demonstration research and care programs through Centers Without Walls where patients with Huntington's disease and allied disorders can be studied, and where full programs of treatment, rehabilitation, counseling, and other services can be developed, evaluated, and demonstrated. Scientists nationwide can collaborate on common problems, sharing common resources.

- Develop new roles for health professionals and paraprofessionals in genetic counseling and psychological counseling.

- Develop new health care personnel to care for patients with chronic disease at home and in intermediate and long-term care facilities.

- Coordinate existing health care services and develop new ones for Huntington's disease families and others with related disorders through the establishment of pilot "ombudsmen" programs in one or more states or regions.

- Conserve existing facilities by converting empty or underused wards or wings of hospitals, state, or Federal institutions to model facilities for the chronic care of patients who need comfortable and hazard-free environments.

The Commission—Mandate and Methods

The Commission for the Control of Huntington's Disease and Its Consequences was established by Public Law 94-63. The Commission was charged to report on the state of the art of research and management of Huntington's disease and to develop a national plan for its control, to be presented to the President and the Congress following a year's deliberations.

The Commission was composed of six professional and three lay persons chosen for their knowledge and personal involvement with Huntington's disease, as well as their interest in public health problems. To carry out its mandate the Commission appointed 15 work groups in the fields of biomedical research and social management. These groups reported on the state of the art of research, care, and treatment of Huntington's disease. They gathered new data, advised on promising leads, and made general and specific recommendations for the National Plan. Guidance and information were also elicited from world authorities on Huntington's disease, leading neuroscientists, geneticists, lawyers, educators, psychologists, and other professional and lay persons.

The Commission held hearings in 11 cities chosen to represent urban and rural regions throughout the country. Almost 2,000 persons, Huntington's disease patients and family members, government officials, health care providers, and others testified in person or wrote to the Commission to describe their personal experiences and problems, and to make recommendations.

Additional sources of information included position papers and special studies on health services, clinical care, legal rights, new drug development and psychological training. Special assistance was provided by the National Institute of Neurological and Communicative Disorders and Stroke, the National Institute of General Medical Sciences, the National Institute of Mental Health and other Government agencies involved in health matters. Advice and aid were also given by the Pan American Health Organization and the Health Services Research Center at the University of California at Los Angeles. The Commission is especially indebted to the Huntington's disease health voluntary organizations: the Committee to Combat Huntington's Disease, the Hereditary Disease Foundation, the Huntington's Chorea Foundation, and the National Huntington's Disease Association, and to the many patients and families these organizations represent.

The Disease and Its Consequences

The fear of losing one's mind and the fear of losing control over one's body are among the most profound fears known to mankind. Both losses occur in Huntington's disease, a hereditary brain disorder which begins insidiously, usually in middle age, and progresses inexorably to death 10 or 20 years later.

Huntington's disease (formerly called Huntington's chorea) causes extensive physical and mental changes. The physical signs may appear only as some clumsiness or nervous fidgeting at first. As the disease proceeds, all parts of the body writhe, twist, and turn in incessant, uncontrollable movement. At times the patient's lurching movements and precarious gait have a bizarre, dance-like appearance, called *chorea.* Like choreography, it derives from the Greek word for dance.

The mental changes in Huntington's disease are also insidious and devastating, leading to personality disintegration and loss of all mental faculties. Patients may at first be forgetful, irritable, violent, or excitable; some withdraw into total apathy or depression. By the end of the illness patients will develop a full-blown dementia, with major losses in memory, thinking, and reasoning. Many patients will have lost the capacity to speak before this occurs. Severe emotional disturbances, delusions, hallucinations and other schizophrenic-like symptoms, or chronic and often profound suicidal depressions frequently accompany the physical and intellectual changes.

Inheritance Pattern

Huntington's disease is an autosomal dominant disorder. "Autosomal" means that it strikes men and women equally; "dominant" means that each child of an affected parent has a 50 percent chance of inheriting the disorder. The disease does not skip generations, passing over one generation to attack the next. The lucky 50 percent who do not inherit the gene for Huntington's disease break the chain of the illness. Their descendants are free of the gene forever. Those who do inherit the gene will inevitably develop the disease. The gene is said to be "completely penetrant" and will eventually exert its effect—this means that the disease will always appear in those who carry the gene.

No Early Detection Test

At present, there is no early diagnostic test which can distinguish carriers of the Huntington's disease gene from noncarriers. There is no way of knowing who will develop the disease until symptoms appear, usually in the thirties or forties. By that time, the disease has usually been passed on to the next generation. Those who are "at risk" for the disease—who have one parent affected by the illness—must make crucial decisions about marriage, family, and career in total uncertainty.

Having worked in the medical field for twenty odd years, in Australia and here, I feel that Huntington's chorea is the most debilitating disease I have ever seen. Being powerless to help nearly kills me.

Age of Onset

A particularly severe form of Huntington's disease affects young people before the age of 20, but the majority of patients develop symptoms in the middle years. The probability that the disease will appear declines after age 45, but some patients still develop symptoms in their seventies or eighties. It is a very long and wearing wait before a person can feel totally free of the disease.

The precise age of onset of Huntington's disease is difficult to determine because of the insidious nature of the early signs. Subtle mental and physical changes may precede the more dramatic symptoms by as much as 10 to 25 years. There is considerable variation in the age of diagnosis, in the range and severity of symptoms, and in the duration of illness.

Course of the Disease

What never varies is the final outcome. All symptoms worsen. Higher mental processes fade and personality becomes more unstable.

The minor clumsiness or stumble, the restless movement disguised as a pat of the hair or a crossing of the arms become full-scale limb movements in flailing activity through every waking hour. Facial expression distorts into grimaces; speech slurs beyond comprehension and then ceases altogether. Chewing and swallowing grow difficult and choking is a constant hazard. Bowel and bladder control are lost. To protect against falls and self-injury, the patient may have to wear padded clothing and be strapped to a bed or wheelchair. Death comes at the end of 10 to 20 years of unrelenting destruction of the nervous system. The patient, exhausted, emaciated, succumbs to heart failure, pneumonia, or other infections . . . or else dies from choking.

No Effective Treatment

There are no effective therapies that will work throughout the course of the disease. Some of the major tranquilizing drugs used in treating schizophrenia are of some help in calming milder choreic movements. Other drugs may relieve anxiety or depression. But side effects are unpleasant and dose rates are often so high that the patient may prefer the disease to the torporous or apathetic state the drugs can produce. And no drug can forestall the progression of the disease. Unlike some cancers, unlike multiple sclerosis and many other diseases, there are no states of remission in Huntington's disease. The bleak fate that now awaits the Huntington's disease patient can be summed up in one sentence from a recent text in neurology:

> Institutional care is required at the late stage unless the subject has already committed suicide or is bedridden with chorea.

The high suicide rate among Huntington's disease patients is well known. Some authorities estimate that it may be as much as seven times the national suicide rate—1.3 percent of all deaths in the United States. Not surprisingly, it is also high among the at-risk group.

At-Risk Experience

Individuals at risk live in a state of uncertainty which imposes a heavy psychological burden. Not only must they bear witness to the painful decline of a parent; they must carry the burden of fear and anxiety that some day the same thing may happen to them. They are unable to plan ahead, to prepare for college, careers, or for love, marriage, and parenthood without apprehension and doubt. They are unable to pass each day without the constant watching for symptoms—the glass accidentally knocked over, a trip on the rug, or just a feeling of depression—all are seen as frightening omens of things to come. Persons at risk feel compelled to conceal their status so they do not jeopardize their jobs, their eligibility for life insurance, medical plans or pensions, or decrease their desirability as marriage partners.

Now the daily wondering and agony — every time I'm a bit nervous, or dropping things, or forgetting things, or accidentally stumbling, or not being able to type as well as I could; it's a relentless watching and waiting, feeling every day that I surely must have it! And, of course, this makes me more nervous and jumpy.

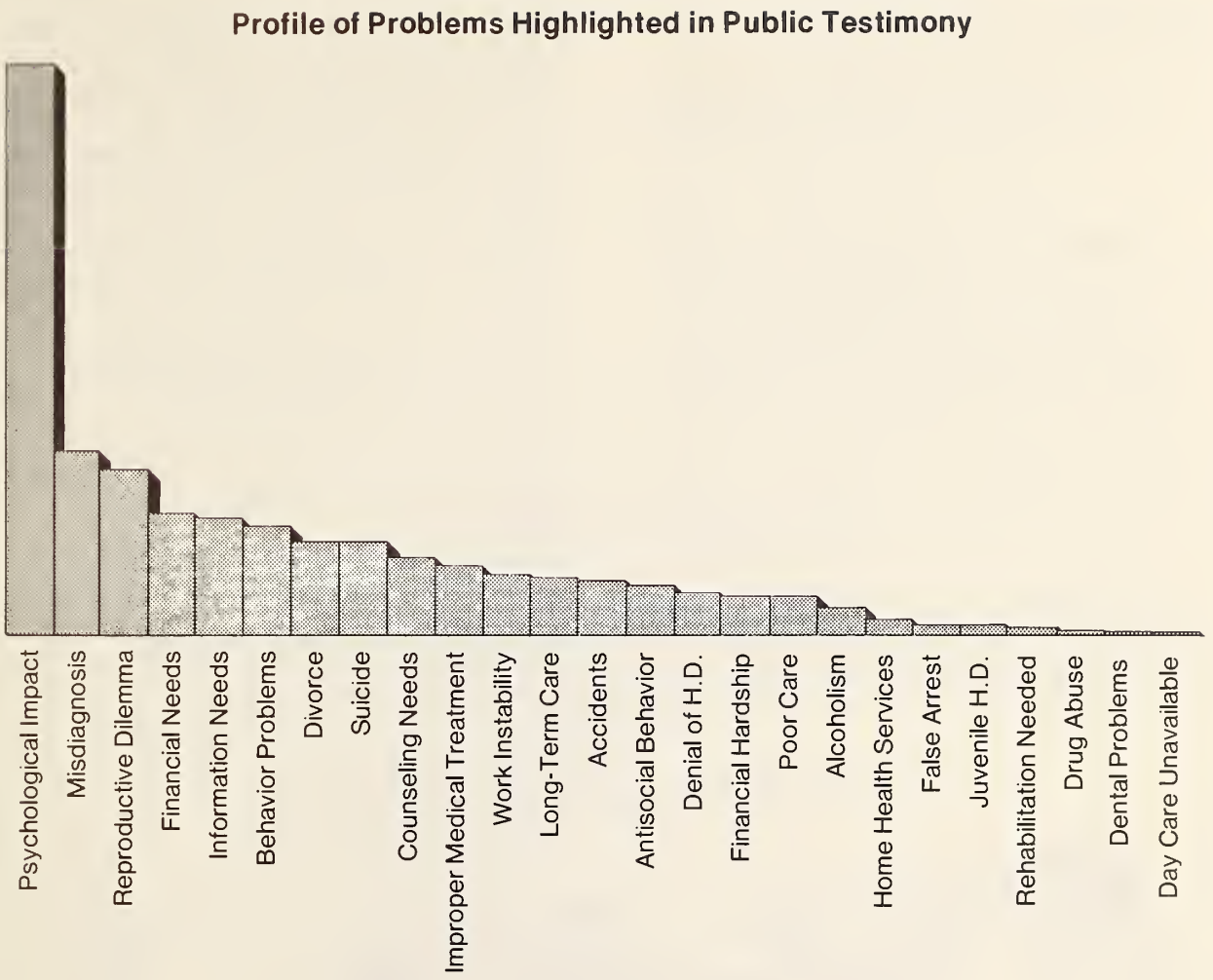
The Need for a Presymptomatic Test

A safe and reliable presymptomatic test would at least relieve the burden for half this group. They would be free—free to be clumsy, to trip, to drop things, to get angry, to cry and feel sad—without the constant fear that these normal human experiences mark the onset of the disease. They would be free to love without fear of imposing a future burden, to marry and to have children secure in the knowledge that their children would be safe from the disease.

If a presymptomatic test were effective prenatally, it could lead to the gradual disappearance of the disease from the population. This has begun to happen in the case of the fatal hereditary brain disorder of young children, Tay-Sachs disease.

For those for whom the test proved positive, however, in the absence of effective treatment there are very real dangers: despair, withdrawal, abandonment, even suicide. Family relationships could be strained to the breaking point if some members were identified as carriers and others were not. The heartbreak of parents in raising healthy, normal children whom they know are destined to be stricken might be too great to bear.

The development of a *reliable* and *safe* predictive test is a long-term process. It will take many years to validate a test because scientists must wait to see who does or does



not develop the disease. In the meantime, those at risk must be protected against learning premature results which could lead to disastrous consequences. People who thought they were free from the disease might have children, only later to develop symptoms. On the other hand, a false positive result would certainly precipitate depression or possibly suicide in a perfectly healthy individual.

Prevalence

Huntington's disease has been found in all races and countries in the world. While the identification of its hereditary nature was made only a little over a hundred years ago, most epidemiologists believe that the disease has existed for centuries. Present estimates of the prevalence rate range from 4 to 7 per 100,000 although recent evidence suggests that it may be as high as 10 in 100,000. Approximately twice this number of patients are at risk for the disorder.

It is difficult to obtain accurate incidence or prevalence rates on Huntington's disease because patients are often misdiagnosed as schizophrenic or as suffering from other neurological diseases. Some families also deny the existence of the disease because of fear, shame, or guilt. Other families break up with the result that patients are abandoned and left to become marginal members of society, out of touch with any medical services. The precise number of patients with Huntington's disease only minimally reflects the number of lives deeply affected. For each patient there is usually a spouse; there are siblings, children, grandchildren, and even great grandchildren whose health and well-being are threatened.

And so, out of all of this, from my grandmother, there are 108 great-grandchildren of hers. Now, this does not show the great, great-grandchildren, which there would be 16 in our family right here. So, as you can see, just one family has exploded, you might say. And we need to have research and know whether they are at risk or not at risk.

The Vicious Spiral of Costs

The true cost of Huntington's disease—financially, legally, socially, and emotionally—must be measured in terms of its impact on the whole family as well as on the entire society.

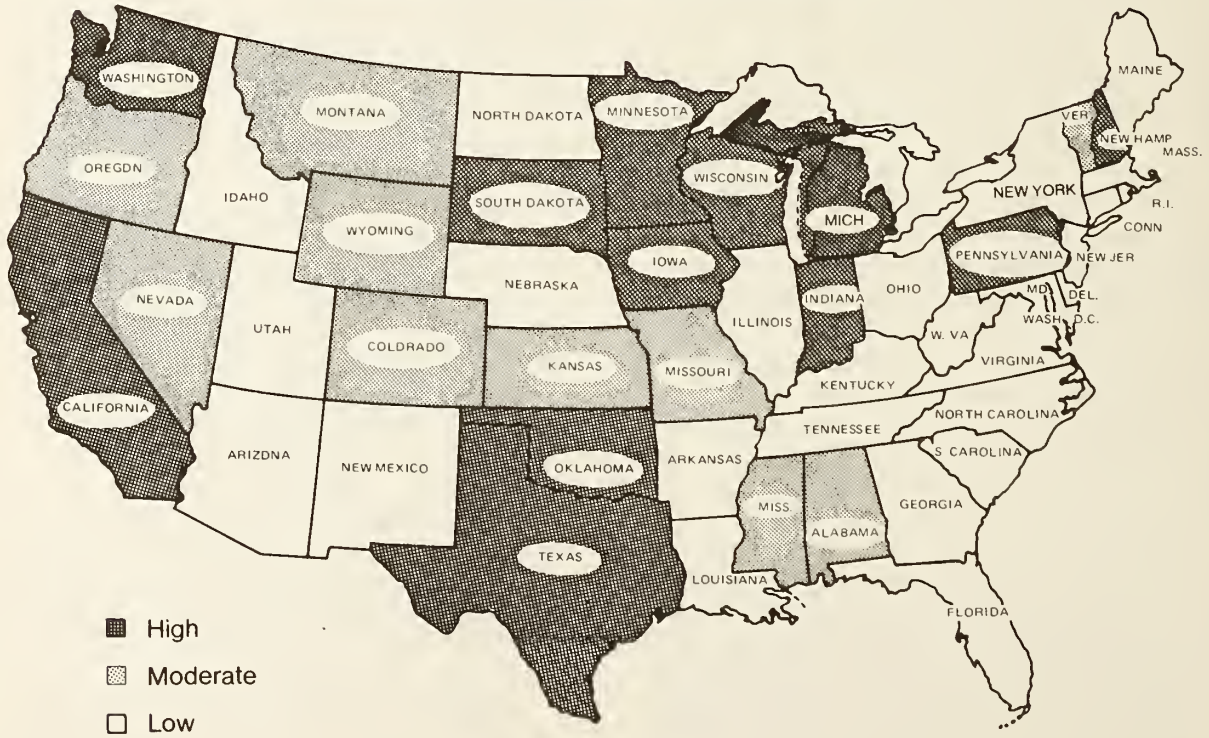
This statement can be made of many diseases, but it is particularly true of late onset hereditary illnesses such as Huntington's disease. Several generations can be suffering from the disorder at the same time. A parent may die just as the child is being diagnosed. The cycle repeats in its inexorable fashion without ever giving the family a chance to recuperate. Not only is it psychologically devastating for the offspring to see the end stage of the disease just as they are confronting their own slow decline, but these cycles generally demolish a family financially as well. There can be no way to save for the next generation in jeopardy.

Because the disease most often shows up in middle life it almost invariably affects family income and life style. If the patient is the breadwinner, a spouse may have to assume that role. Inevitably income drops while medical costs mount in a vicious spiral that affects the mental and physical well-being of all members of the family. Children may have to drop out of school because there is no one else who can stay at home to attend the ailing parent.

In a travesty of care, couples have been advised to divorce if a spouse has Huntington's disease. This would avoid financial ruin and perhaps allow the couple to save for the future care of those at risk. The government would then assume the responsibility for care of the indigent patient.

Huntington's disease is a "downwardly mobile" disease fraught with fear, shame, guilt, and an overwhelming financial burden. That burden may be borne by the family at first, but inevitably it becomes society's

Huntington's Disease Reported Mortality



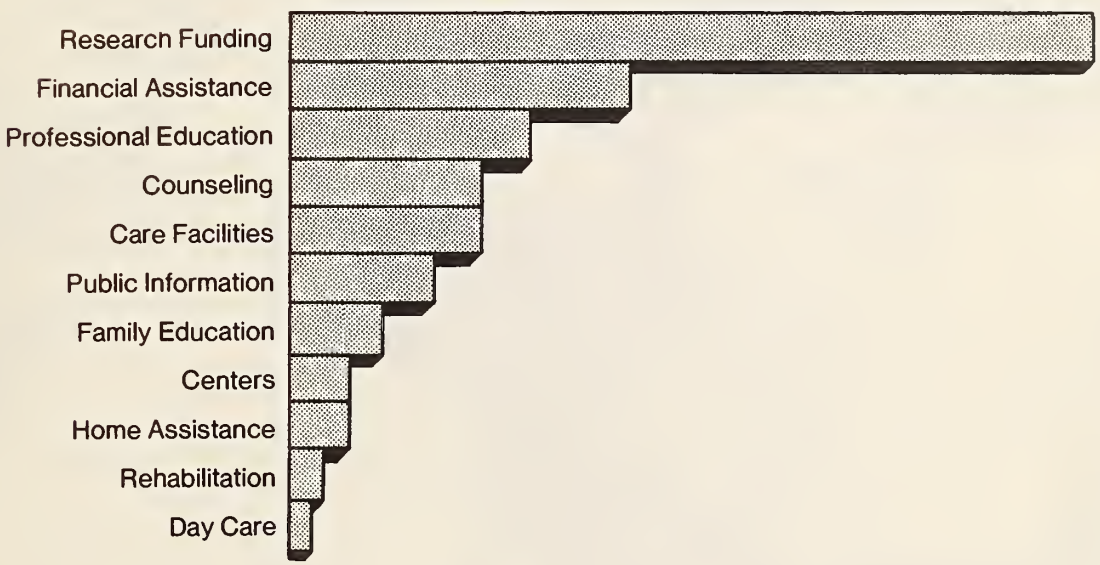
*Information based on preliminary information derived from the Work Group Report on Epidemiology, Biostatistics, and Population Genetics.

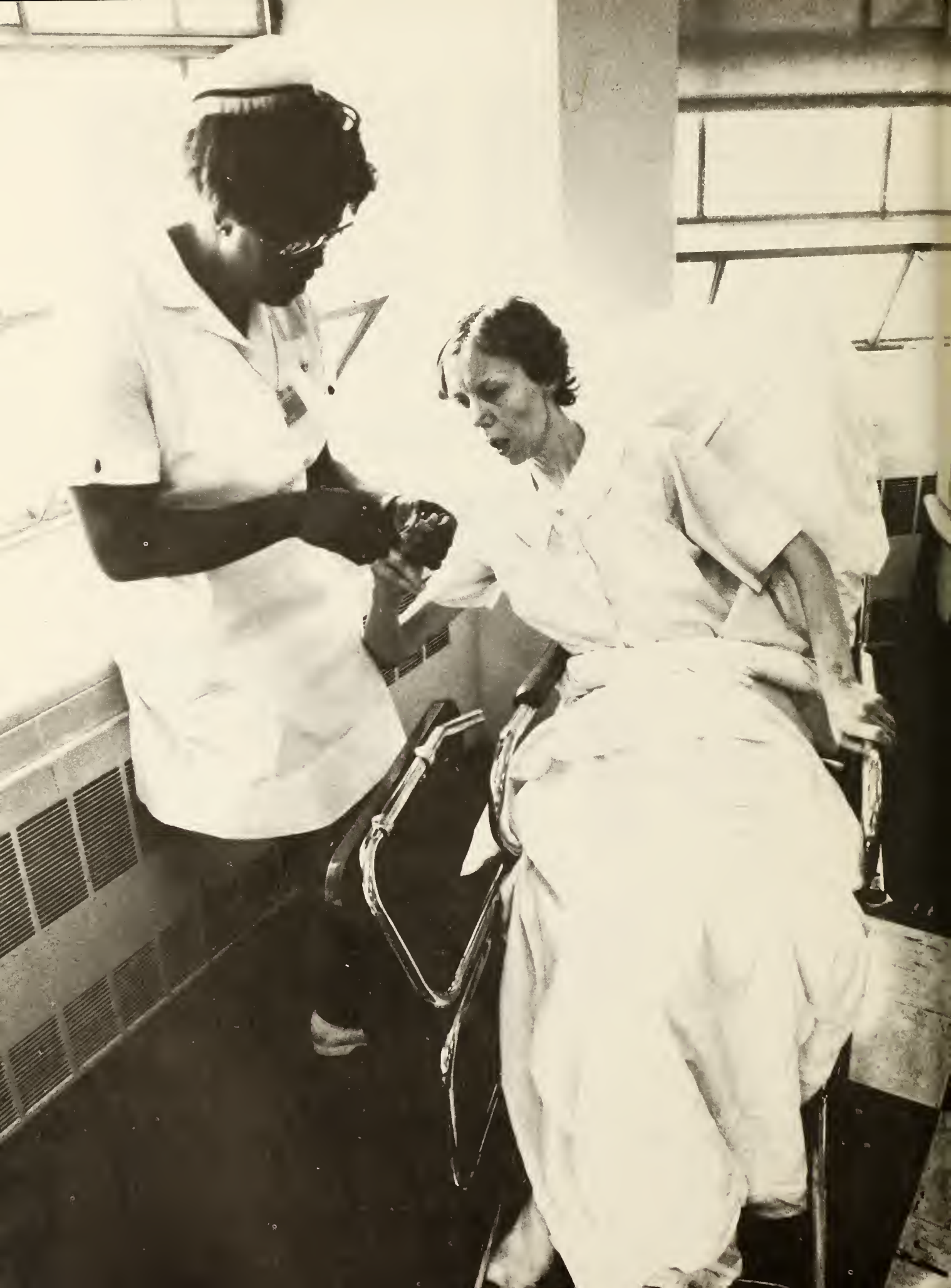
problem. Along with the loss of productive labor and taxable income, society must absorb the cost of chronic medical care and hospitalization—not just for one patient with Huntington's disease and the immediate family, but for a pyramiding population of relatives now and in generations to come. (See p. 56 for specific costs.)

Truth of this disease is stark. Death of a Huntington's patient is perhaps the least tragic aspect of the disorder; death is almost a kindness. The emotional agony of years of uncertainty and threat which pervades every aspect of life, the inexorable unfolding of a slowly dementing, grotesquely deforming disease, the bankrupting cost of health care extending over decades, the gradual loss of all physical and mental control represent a waking nightmare almost beyond comprehension.

> Probably that you die is the part that scares me the least. It's almost more human than the part where you live. It's an awful thing to look at your kids and wonder if some day they're going to look at you like some kind of monster.

Profile of Recommendations Taken from Public Testimony





Huntington's Disease as a Prototype

A Prototype for Research

> Nature has a way of revealing her secrets through one rare disorder.
>
> Irving Kopin, M.D., 1976

The wide range of mental and physical symptoms of Huntington's disease encompasses the collective misery of major hereditary, neurological, and psychiatric disorders—each of which alone is serious, chronic, or fatal. Tragic as this is for patients and families, it allows researchers to study and explore Huntington's disease as a prototype for many.

Neurological Disorders

Huntington's disease produces disturbances in movement, thinking, and feeling. Scientists now believe that the constant writhing and twisting of the Huntington's disease patient, the inability to control or stop the movements, is related to the loss of important brain chemicals which inhibit nerve action. This line of reasoning grew out of discoveries made about Parkinson's disease, a related neurologic disease. Parkinson's disease patients show rigidity and tremor. Studies of the brains of these patients revealed a low level of dopamine, a chemical which facilitates or excites nerve action. This led to the development of the drug L-dopa, which helps restore dopamine levels to normal and so relieves the symptoms of Parkinsonism in many patients. Subsequent studies of the brains of Huntington's disease patients showed lowered levels of a substance which inhibits nerve action—gamma-aminobutyric acid (GABA). The search is now on to find a drug which can restore the level of this substance to normal with the hope that it would provide symptomatic relief. Other neurotransmitters have also been discovered to be altered in Huntington's disease, opening new realms of exploration for a treatment.

Research on Parkinsonism led to an insight into Huntington's disease. Research on Huntington's disease may shed light on Sydenham's chorea, hereditary tremor, Gilles de la Tourette's syndrome, narcolepsy, the cerebellar ataxias, multiple sclerosis, amyotrophic lateral sclerosis, and even Parkinson's disease—the cause of which is still unknown. It is impossible to predict exactly how a better understanding of the brain will lead to new treatments or cures for disease, or which disease will be most affected. But it is certain that without such knowledge, thousands will continue to suffer and die.

Hereditary Diseases

There are over 2,300 genetic diseases that have been described, 500 of which are neurological. Huntington's disease belongs to the group of nearly 1,000 *dominant* gene diseases. These include achondroplasia (dwarfism), some cerebellar ataxias, some forms of muscular dystrophy, familial hypercholesterolemia (a condition predisposing to heart disease), and polyposis (a disorder that leads to cancer of the colon). Many heritable traits—such as blood type or certain diseases—are determined by two genes. One gene is contributed by the father; one by the mother. A "dominant" hereditary disorder is one which can be produced if only one gene of a pair is defective. The corresponding gene may be normal. "Recessive" disorders are ones in which both genes must be defective.

Considerable progress has been made in the understanding of recessive disorders. The symptoms of such diseases may vary widely, but there are similarities in the way the defective genes produce their damaging effects. When the mechanism underlying the recessive brain disorder Tay-Sachs disease was clarified, for example, it led to an understanding of a whole group of recessive disorders. Scientists knew what to look for and how to look. This greatly simplified their efforts and accelerated research which led to ways of detecting carriers of recessive disease. Studies indicate that about 30 percent of hospital

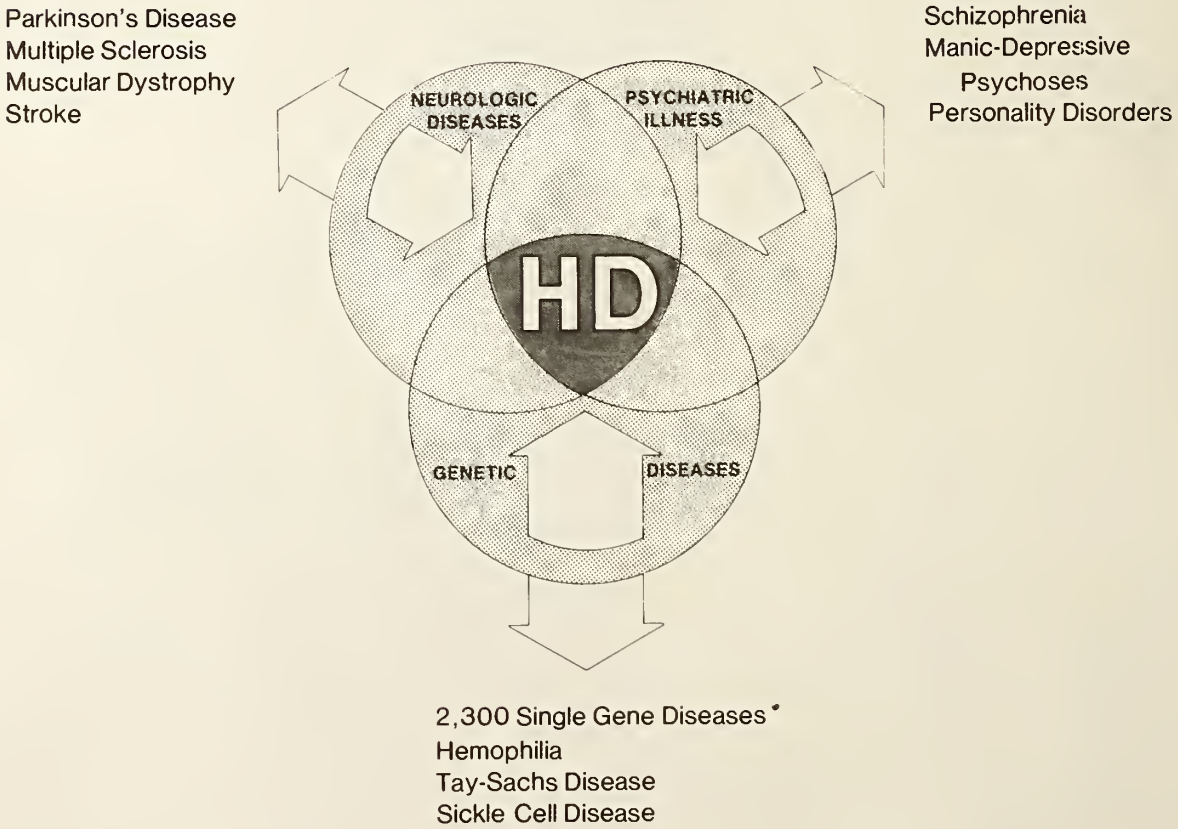
ized children have diseases of genetic origin, 40 percent of all infant mortality results from genetic factors, and 80 percent of all mental retardation in the United States is genetically related. It is now possible, however, to diagnose some 60 serious genetic disorders before birth through a process known as amniocentesis, and the list is growing yearly. Unfortunately, no dominant hereditary disease can be detected prenatally.

Far less is known about dominant genetic disorders. Why should one gene alone be so powerful as to cause disease? What part of the cell does a dominant gene affect? Are its effects widespread, affecting many cells in many organs of the body, or are they more specific, confined only to the brain, for example? As with recessive disorders, once one disease is understood, it opens the door to explaining many others. Understanding how the gene for Huntington's disease produces its damaging effect would, in all likelihood, be such an opening wedge.

Psychiatric Illnesses

Another group of brain disorders is characterized by emotional disturbance and confusion of thought. Schizophrenia and manic-depressive psychosis are the most widespread of these brain disorders, with symptoms very similar to the changes seen in Huntington's disease. Patients with Huntington's disease often suffer disabling personality changes; they show extremes of emotions: excitability, hostility, violence, and depression. They may experience hallucinations, delusions, and other psychological symptoms as much as 5 to 25 years before the movement disturbances begin. Psychiatric disturbance is the first indication of the onset of Huntington's disease in almost 50 percent of cases according to a recent Swedish study. Many patients experience chronic moderate depression. This symptom is especially disturbing to persons at risk who worry that bouts of depression, which are common in everyone's life, are in their case

Huntington's Disease as a Prototype for Research and Treatment of Other Neurologic, Psychiatric and Genetic Diseases



premonitory symptoms of Huntington's disease. Very frequently Huntington's disease patients are misdiagnosed as schizophrenics and treated as mental patients with no understanding that the disease is inherited and that they should receive genetic counseling.

The advent of drugs to treat schizophrenic and manic-depressive patients has added weight to the belief that the major psychiatric cripplers have a biochemical component at some level. Lithium carbonate is effective in relieving manic-depressive psychosis and there are other drugs that can significantly improve many patients with schizophrenia. Curiously enough, the only medications which provide even minimal relief for the constant movements of Huntington's disease are the drugs used routinely to treat schizophrenia. Perhaps there is a biochemical link, deep within the brain, connecting the two diseases.

There is also increasing evidence for a hereditary factor in schizophrenia and manic-depressive psychosis, although the inheritance pattern is not clear-cut. Both psychiatric diseases are considered polygenic, that is, they involve the complex interactions of several genes in addition to environmental factors. Like Huntington's disease, schizophrenia usually strikes adults. The age of onset is in the late teens and twenties. Schizophrenia is more common than Huntington's disease. It affects approximately 1 person in 100 while Huntington's disease may affect 1 in 10,000. But the pattern of inheritance of Huntington's disease is simpler. It is a single gene disease rather than polygenic, and follows strict rules of inheritance. This makes it a very useful model for psychiatric illness. Investigators can study an easily identifiable volunteer population at risk for Huntington's disease before symptoms appear, and as the disease unfolds in some patients. Moreover, since the symptoms of Huntington's disease stem more closely from the gene itself, some of the complex environmental variables that make schizophrenia research so difficult are eliminated. Perhaps a clue to some of the major mental illnesses of our time lies in solving the puzzle of Huntington's disease.

The Aging Process

It is known that the gene for Huntington's disease leads to early death of certain nerve cells in the brain. Since the loss of brain cells occurs normally and steadily in the course of maturity, Huntington's disease is sometimes described as a kind of rapid and premature aging. Physically, Huntington's disease patients also appear to age rapidly so that a 45-year-old patient may look like an ailing person of 60 or over.

Throughout the life cycle inner "biological clocks," controlled by genes and interacting with the environment, regulate when the normal milestones of growth occur: from walking and talking, to sexual maturity, to the wrinkled skin and brittle bones and other changes associated with age. Genetic factors undoubtedly make some people more vulnerable to disorders of aging, so that they are more prone to the loss of intellectual and physical ability, becoming senile and feeble. These biological clocks are as little understood as the genetic clock that controls the timing of Huntington's disease: an individual looks healthy and is active until middle age and then, mysteriously, a slow decline begins. If the Huntington's disease gene is present at birth, why does it wait until middle age to "turn on"? Or does it? Perhaps the gene begins its destructive process even before birth, but is unmasked only when the effects of normal aging accumulate and other cells begin to slow down and die.

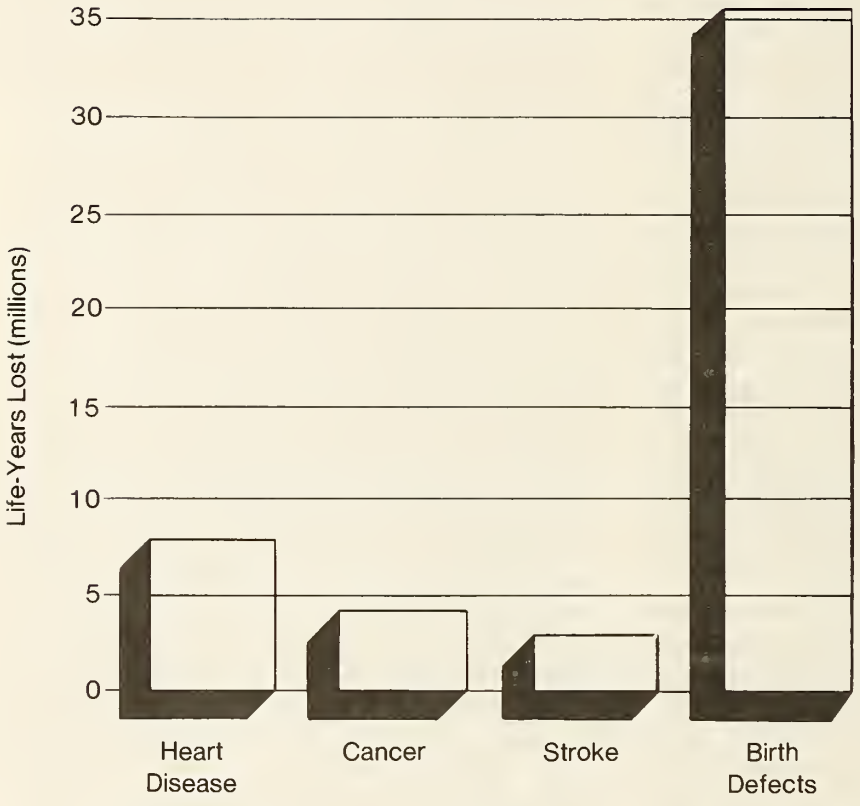
Senility

One of the most fearsome diseases of the aged is senility; for, when far advanced, it entails dementia—the loss of the intellectual faculties of thinking, reasoning, memory, judgment, and the like. Dementia is the most terrifying consequence of Huntington's disease. People at risk and in the early and intermediate stages of the disease uniformly dread the prospect of "losing their mind" and "becoming a vegetable."

Huntington's disease is classified as a "*presenile* dementia," which means that the loss of intellectual abilities occurs before old age.

It is estimated that some 8.4 percent of the population suffer from severe to mild dementia, including such diseases as Alzheimer's and Niemann-Pick disease. There are one million

Loss of Life-Years Resulting from Genetic Disorders



Birth defects, of which about 80 percent are thought to be genetic in origin, are a leading cause of infant mortality. As such, when measured in terms of the normal life expectancy, they are seen to claim 4.5 times as many life-years as heart disease, 8 times as many as cancer, and 10 times as many as stroke.

Source: U.S. Department of Health, Education, and Welfare, *What Are the Facts About Genetic Disease?*, DHEW Pub. No. (NIH) 76-370, Figure 5.

severely demented and another three million mildly to moderately demented persons in the country today. Severe senile dementia ranks as the fourth or fifth leading cause of death in the U.S.

At least two-thirds of cases of senile dementia have classical Alzheimer's disease. It has been suggested that there is a strong genetic predisposition to developing this type of severe dementia.

Despite its prevalence, there is little research underway on dementia. The combined resources of the National Institute of Neurological and Communicative Disorders and Stroke (NINCDS), the National Institute on Aging (NIA), and the National Institute of Mental Health (NIMH) support a mere 12 significant research grants in this area. Understanding the cellular biology of Huntington's disease might reveal a common pattern in all dementias leading to a therapy which would allow millions to live out their lives in full possession of their faculties.

Shared Interests and Shared Concerns

The Commission does not claim that because Huntington's disease is a prototype for so many serious disorders it should be studied exclusively. The serendipitous nature of scientific discovery argues against such a view. As the Nobel laureate, Carleton Gajdusek, remarks, "It came as a surprise . . . to discover that certain degenerative disorders were virus infections."

To allow for surprise, for the painstaking work in one field of study that suddenly illuminates another, the Commission believes that research support should be basic and broad. Research on Huntington's disease itself is important, but so too is research on the basic organization of the brain and research on other disorders. For often as not, the breakthrough for one disease emerges from studies of other related disorders or even from seemingly unrelated areas of science.

Huntington's Disease as a Part of a Whole

Research is not the only area in which Huntington's disease has bonds with other disorders. The Commission learned throughout the year of the desperate need for certain services—financial relief, appropriate long-term care facilities, mental health services, rehabilitation, education and counseling programs, and legal protections. Little effort was required to see that these needs were shared with others, particularly those with chronic disease.

There is a gradual change taking place in attitudes toward health which puts a new emphasis on caring rather than curing. Vast sums of money and energies have been invested in sophisticated treatments of acute or infectious illness. Accomplishments have been so great that they have changed the nature of medical practice. The populations in the country today with the most pressing needs are the chronically ill who suffer from lingering, slowly deteriorating conditions. For these populations, the medical model, focusing on the immediate, acute, episodic needs of the patient must be supplemented by an equal concern for the maintenance of the patient at an optimal level of functioning in social, professional, and family life. Rehabilitative and social service therapies are assuming prominence as part of a comprehensive health care model. The increasing numbers of elderly in our population, many of whom have chronic conditions, may serve to catalyze this shift. Dr. Stanley Brody of the University of Pennsylvania describes needs of the elderly which are equally applicable to the chronically neurologically impaired of any age:

> While the aged have need for acute medical care, their major requirement is in the continuum of services for the chronically disabled that will enable them to function optimally. Any health system that continues to be limited to a disease orientation will not meet the increasing needs of the aging community. Medical services must take their place as a part—and only part—of the continuum of health care.

The Commission identified numerous gaps in the continuum of health care which leave many patients unserved.

The Middle Income Financial Crisis

Comprehensive health care is available and accessible primarily to the very rich or to the indigent. Middle income citizens find that either services do not exist for them or that, over the course of a chronic disorder, they must become indigent to receive financial relief or medical benefits. Social Security regulations which provide for disability insurance and supplemental income payments create an artificial "notch." Earnings above the notch make a person ineligible for any medical relief benefits, even for catastrophic long-term disease. Earnings below the notch preclude a decent job. Medical benefit eligibility requirements are a disincentive to work.

Often, the ground rules of insurance or government-supported programs dictate the course of treatment to the detriment of the patient and at increased cost. For example, if only hospitalization is covered, the patient can go to a hospital at high cost but can't go to a convalescent hospital at lower cost or have home care at perhaps even lower cost—also a more desirable situation.

In families with hereditary disease, there is no way to save to provide financial protection for at-risk children. Financial relief is critical to individuals and families struggling with chronic disability. *Faced with a choice between poverty or divorce—leaving the patient indigent and therefore eligible for benefits—many families splinter.*

Lack of Adequate Long-Term Care

Expenditures on long-term care are expected to increase from approximately $13 billion in 1975 to upwards of $31 billion in 1980. Federal spending under existing programs would be about $7.5 billion in 1980.

Between 1960 and 1973, expenditures on nursing home facilities, including staff and care, increased 1,400 percent.

The range of demand for long-term care is estimated to increase from between 5 and 10 million persons in 1975 to between 7 and 12 million in 1985.

Yet it has been estimated that as many as 400,000 residents in nursing homes do not need to be institutionalized. Some 20 to 40 percent of the nursing home population could be cared for at less intensive levels were adequate community-based care available.

As alternatives to institutional care—or no care at all—sheltered living arrangements, congregate housing, home health care, respite care, and day care are sadly lacking.

Nursing home abuses—leading to death and injury on occasion—are widespread. Estimates of the number of substandard homes (those in violation of one or more standards causing a life-threatening situation) vary from 30 to 80 percent. Inspection and enforcement regulations are largely ineffectual.

Special precautions must be taken to insure that the chronically disabled—be they young or old—are guaranteed their human rights and protections and that they are provided with the optimal level of care needed for their disability.

My mother had not worked outside of the home since before she married and so she was not eligible for Social Security, or any other benefits. This placed the full burden of a nursing home costing over $500 a month on my dad who was also trying to put the two children still at home through school.

Fragmentation of Services

A bewildering array of services and programs administered by a multitude of agencies and organizations at the local, state, and Federal level defeats rational and coordinated service delivery. Individuals with similar needs are treated differently depending on age, income level, type of disability, and state of residence. There must be coordination of programs and immediate and simplified access to eligibility information.

Neglect of Mental Health Needs

It is common knowledge that emotional disturbance can contribute to the production of physical illness. The emotional distress of persons trying to cope with severe or terminal disease or the threat of disease, however, is often ignored. Because patients are considered to have a physical ailment, their psychological needs are neglected. Counseling interventions at the time of diagnosis and subsequently over the course of an illness may prevent or diminish emotional disturbance both in patients with organic disease and in family members who are also vulnerable to emotional disturbance.

Denial of Rehabilitation Services

Current laws are interpreted to provide rehabilitation services and vocational training almost exclusively to persons with remediable or stabilized conditions, even though they may be severely handicapped. Persons with slowly progressive and eventually fatal illnesses such as Huntington's disease, multiple sclerosis, and Friedreich's ataxia have been disallowed. Many patients can continue productive lives for many years after illness begins, even if former pursuits are not possible. The emphasis should be on the rehabilitation of an individual to a maximum degree of functioning—whether work for remuneration is possible or not.

Opening the Doors for All in Need

Young and old patients with chronic and progressive disease too often are condemned to isolation, loneliness, boredom, and fear—subsisting on the margins of society. This is particularly true if their mental faculties are involved. Persons with Huntington's disease and other chronic progressive neurologic disease have all too frequently been shunned by the very caretakers who should give solace—physicians, nurses, mental health professionals, family, and the public. They frustrate the medical imperative to cure. They force reconsideration of human values. A new outlook is just beginning in this country—one that invites participation of the handicapped, the neurologically impaired, and the elderly in the mainstream of society. Even death is gradually being treated with less fear. This outlook is very new and fragile and must be encouraged.

How can I adequately convey to you what the pressure of being threatened by Huntington's disease is like? Let me try by telling you about two possessions I keep.

One is a picture, a photograph from a newspaper clipping. It is one of those rare photographs that captures a scene so pathetic, so utterly piteous that you try to tell yourself that such a thing cannot possibly be real. But it is, undeniably real. It shows a woman in her forties. She is strapped into a wheelchair. She is obviously out of control; legs flung high, knees to her chin, feet high over her head, hands clutching. Her face is a study in anguish; lips stretched, teeth bared, jaws pulled wide and strangely askew, tongue out-thrust, eyebrows leaping. Her body is emaciated to the point where every bone and tendon is visible through her clinging skin. She is dressed in a diaper. The caption to the photograph tells us that the woman is a victim of Huntington's disease. I keep it as a ready and infallible reminder of what Huntington's disease really has to offer me should it become my lot.

My other possession is a pistol, a .38 caliber police special. It is my insurance that I will never end up like the lady in the photograph.

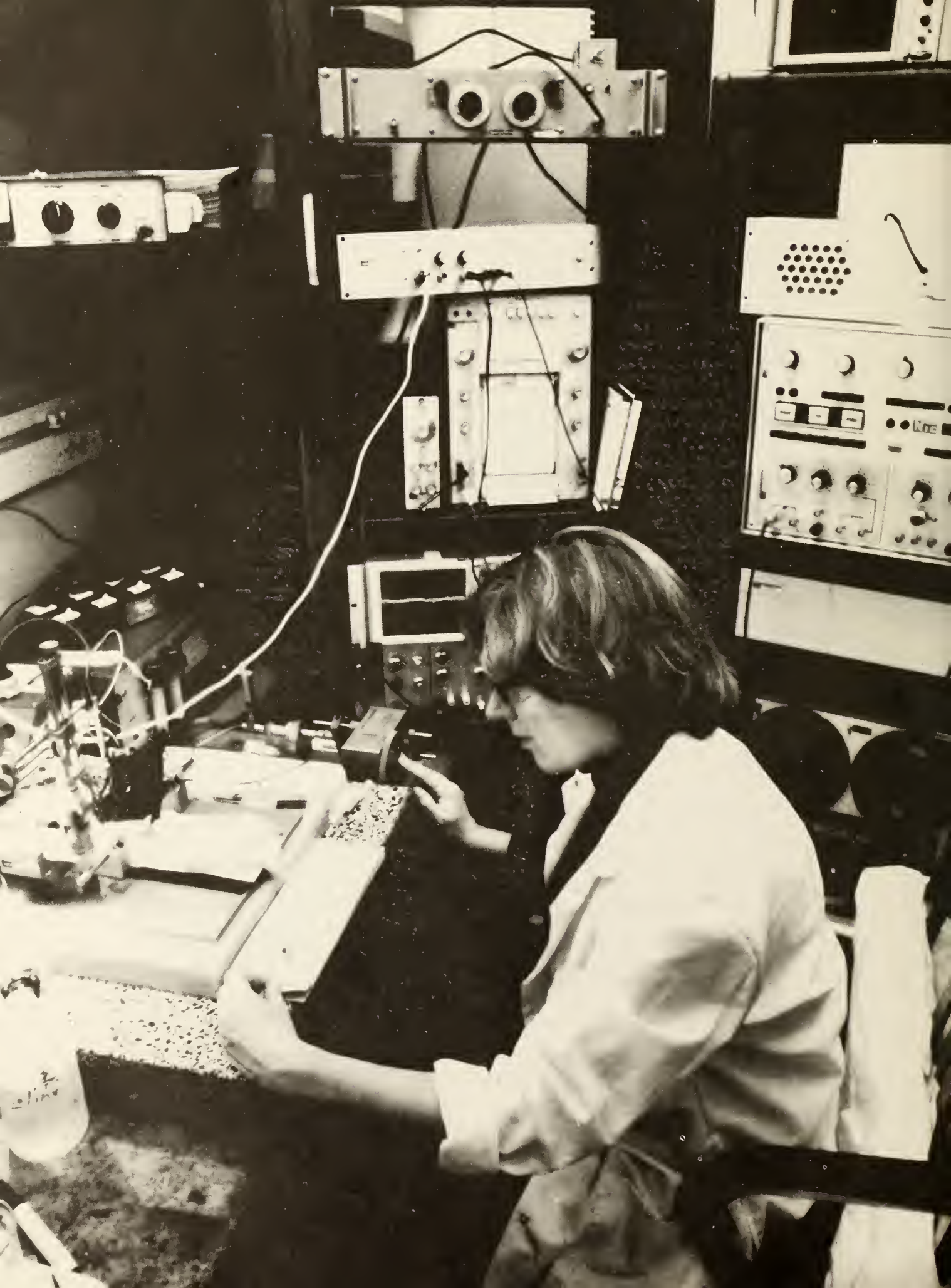
One aspect worth mentioning is the memory I have to live with; the memory of my childhood home being turned into a complete shambles by the mother I loved growing steadily more crazy and debilitated. I can still feel the horror that gripped me the first time I saw my mother's urine run down her leg and across the kitchen floor. My heart sinks to recall all the fighting, the yelling, and the crying. And there was the agonizing over the painful decision finally to have her committed. All of this due to Huntington's disease, and at the time none of us even knew why. It is a bitter memory to live with.

I am now thirty-two years old. I first learned of Huntington's disease when I was twenty-three. That means that for nine years now I have been watching every move I make; checking and testing myself to see if the coordination and memory are still there, wondering if each mistake, each spilled glass of orange juice, each show of anger might be the first sign.

When my brother or I do something clumsy the joke is, "Get away from me, buddy. You've got Huntington's!" but inside no one is laughing. Being at risk is dying a little inside each time you drop a spoon. Being at risk is looking at your brothers and sisters, wondering which one of you will be the first to go. This constant pressure, day in, day out, year after year, takes a heavy toll.

Being at risk creates another set of problems which also must be dealt with. These are the questions about marriage and children. Any woman who accepts me must accept my 50-50 chance of having the disease. I have seen the absolute hell Huntington's disease victims can create for their spouses, and the thought of myself doing that to one I love fills me with dread. And how does this sound to a prospective fiancee? "I love you, Dear, but if you marry me you take a 50-50 chance that I'll have Huntington's disease, and if that happens the MOST I can promise is that I will try my best to blow my brains out." I feel at times that being at risk has taken away my right to love and be loved.

The problems and pressures of being at risk for Huntington's disease are many, and they intertwine themselves inexorably around every aspect of the at-risk person's life. Obviously, the only cure for the at-risk person's situation is the cure for Huntington's disease itself. I feel that the best chance for finding a cure or a preventative for Huntington's disease is through substantial Federal support of primary research.



Research: The Hope for Answers

The Commission recommends that Congress increase appropriations to support basic and clinical research on the nervous system, in genetics, and in other areas relevant to an understanding of Huntington's disease and other chronic genetic, neurological, and psychiatric illnesses and the diseases of aging. These appropriations should support research at appropriate institutes of the National Institutes of Health (NIH) such as the NINCDS, the National Institute of General Medical Sciences (NIGMS), and the NIA, as well as other bureaus, institutes, and divisions of the Federal government such as the NIMH and the Veterans Administration (VA).

Huntington's disease presents challenges in research in two of the most promising disciplines of biomedical science: genetics and the neurosciences. The President's Biomedical Research Panel in its report of April 1976 cited both these disciplines as areas in need of greater emphasis. "The total impact of these [genetic] diseases on public health is enormous." In turning to the neurosciences, the Panel report continued: "Perhaps the ultimate challenge to biomedical research, representing the very pinnacle of our understanding of the human organism, lies in neurobiology: how the brain and nervous system work, how they function in health and disease. . . . This Panel commends neurobiology as a compelling long-range interest worthy of national attention."

Central to the National Plan developed by the Commission is the recommendation for increased support of basic and clinical research on the nervous system, on genetics, and other relevant fields. Only in this way can fundamental understanding of the cause and process of Huntington's disease and other baffling disorders be obtained—as well as information about the normal brain.

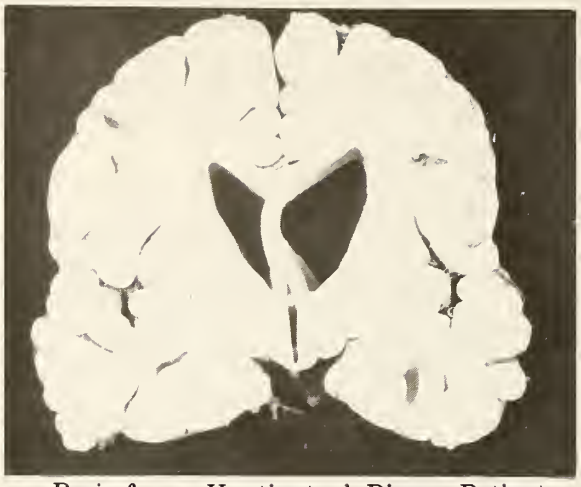
Although targeted research can yield important information, the Commission believes that a less targeted approach and increased support of broad areas of research has a better likelihood of delivering the ultimate answers. The Commission focused its attention on disciplines of general research interest as well as specific research questions.

The Basic Neurosciences

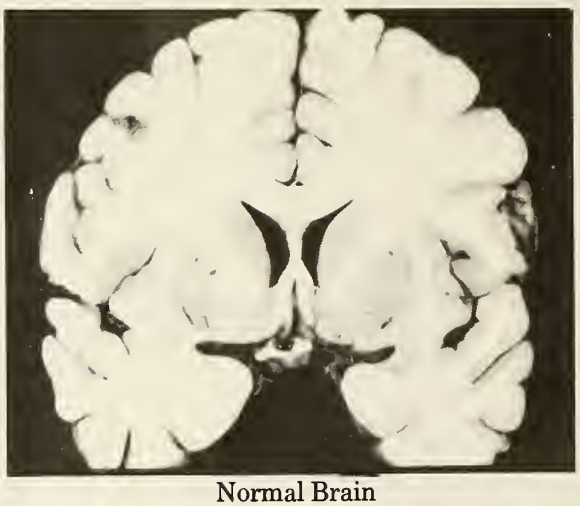
The parts of the brain which are affected in Huntington's disease are particularly intriguing because of the functions which they control. For many years scientists have known that the "gray mantle of the cortex"—the outermost layers of brain—was involved in thinking and memory, and in perception and judgment. These layers of nerve cells (neurons) are more highly developed in man than in any other species and are among the last to develop in the course of evolution. Under the cortex, in an older part of the brain, are the basal ganglia—masses of neurons which, among other things, are involved in control of movement.

Parts of both these critical areas of the brain are progressively destroyed in Huntington's disease. It seems reasonable to surmise that the symptoms of the disease—the progressive loss of mental faculties and the development of chorea—are the outward sign of these inner cell losses.

More recently scientists have established that there are also cell losses in Huntington's disease in other parts of the brain. A challenge to investigators is to discover more about the functions of various parts of the brain by carefully correlating damage to the structures of the brain with clinical symptoms in Huntington's disease patients. This requires careful recording of the patient's clinical history—the excitements, depressions, anger, apathy, extent of movement, loss of intellect, and so forth—and progressive study of cell loss by noninvasive techniques as well as retrieval of brain tissues after death.



Brain from a Huntington's Disease Patient



Normal Brain

Structural and Functional Studies

Establishing the links between brain structure—the anatomy of cells and their interconnections—with function—how human beings think, feel, or act—has long been a major focus for research on the nervous system. Neuroanatomy, neuropathology (the study of diseased nerve tissue), and neurophysiology (the study of how nerve cells function and are connected) are major disciplines in what are now called the "neurosciences."

Many would like to know what brain mechanism underlies memory, why the ability to remember improves in early growth but fades with age, and where thoughts are generated in the brain. It would be equally momentous to know what brain cell activity underlies moods and gives rise to anger, sexual feelings, and joy.

A disorder like Huntington's disease is of special importance in this research since it can cast light on precisely those areas of the brain that are least understood and quintessentially human. So many regions of cortex remain unknown territory that they are still referred to as "silent areas."

I think the essential point is that the time is right for Huntington's disease. This is the basic fact that we have to start with. More new possibilities for research have opened up in the last five years than in the previous 100 with respect to Huntington's disease.

Because Huntington's disease is a slowly progressive brain disorder it offers the opportunity for relating brain changes to behavior over many stages of the disease. For example, the loss of cells in the cortex and basal ganglia that is the hallmark of Huntington's disease is always found in the late stages of the disease. But it is not clear where or how the degenerative process begins. Do cortical or other brain changes occur earlier, later, or at the same time as in the basal ganglia? How many brain areas are involved? Do the changes proceed at different rates? And, if so, could such variations account for the variations in the symptoms individuals develop? Answers to some of these questions may come through computerized tomography and other new techniques as they are explored and evaluated.

These are among the basic questions, questions which should be asked not only for Huntington's disease, but other related neurodegenerative disorders. Findings in Huntington's disease must be compared with those in the psychiatric illnesses, the dyskinesias, ataxias, and other disturbances in the coordination and control of movement. *Basic research is needed on the structural and functional organization of the human brain, especially of the cortex and basal ganglia, to illuminate the mechanisms underlying intellectual faculties, emotion, and the control of movement, and how these mechanisms are affected by disease.*

Neurochemistry

Great advances have been made in understanding the chemistry of the human nervous system in the last few decades. The success of tranquilizing and antidepressant drugs in treating schizophrenia and manic depression—as well as treating milder states of stress or neurosis—intensified interest in the role

played by chemicals produced by the nervous system or by glands under the control of the brain. New discoveries achieved through studying the effects of hallucinogenic drugs and narcotics such as heroin or morphine further clarified the understanding of a family of chemicals now considered to be vital to the normal functioning of the nervous system: the neurotransmitters. Every thought, every gesture, every incoming message or outgoing command of the nervous system depends on the actions of these chemicals in conducting the nerve signal from cell to cell. Sometimes the chemicals facilitate conduction of the message; sometimes they inhibit it.

The discovery that a particular neurotransmitter—dopamine—was deficient in the brains of patients with Parkinson's disease led to the development of the first really effective drug in treating a movement disorder—L-dopa. Now it appears that certain other neurotransmitters are deficient in Huntington's disease. Amounts of GABA—considered one of the most important inhibitory transmitters in the brain—are low in the brains of patients with Huntington's disease in comparison to normal controls. The same is true for chemicals involved in the synthesis of GABA.

All I have to say is I hope research is funded so many questions can be answered on H.D. So that if in the future I get this disease I will not have to kill myself to spare my husband and son. The hell of life they would have to bear with me around would be unbearable to me.

The loss of GABA and other transmitters such as acetylcholine has primed interest in further analyses of Huntington's disease brain tissue to discover the full extent of neurotransmitter changes. The total number of neurotransmitters in the normal brain is unknown. The effects of those which are known are just beginning to be explored. Attention is also focused on the membranes of nerve cells. They contain the receptor sites where the transmitter chemicals released by neighboring cells exert their effects. Abnormalities in receptors have been found in Huntington's disease. Advances in neurochemistry have contributed to the explosion of interest in the neurosciences in the last decade. *Research must be supported to expand studies in these areas, not only to aid the understanding of Huntington's disease but to add to the fundamental store of knowledge of how the human brain works. In turn this will open a window on the understanding of illnesses that are now obscure.*

Neuropharmacology

There is a close relationship between neurochemical studies and neuropharmacology. Many of the drugs currently in use to treat brain disorders directly affect the manufacture, storage, release, or breakdown of neurotransmitters. This includes L-dopa, used in Parkinsonism, as well as the tranquilizers used in treating schizophrenia. Research on drugs that may be effective in Huntington's disease has been directed toward finding ways of restoring normal levels of GABA and influencing the actions of other transmitters in the brains of Huntington's disease patients. *Support for such research and other promising leads to effective treatment for neurodegenerative disease is critical and must be increased.*

Endocrinology

In addition to changes in neurotransmitters in the brains of Huntington's disease patients, there is some evidence that other chemical changes take place involving the relationship between the nervous system and glands of the body. A striking feature of the disease is that many patients appear emaciated in spite of having ravenous appetites. A number of observers have reported that significant numbers of patients show symptoms of carbohydrate abnormalities and diabetes. These changes may not be specifically related to the pathology of Huntington's disease because they also occur in other hereditary diseases of the nervous system. The changes could also be due to a primary defect in glucose metabolism, rather than changes in the nervous system. However, other glandular changes have also been discovered in Huntington's disease patients, such as abnormal amounts of growth hormone produced by the pituitary gland.

The major glands of the body are influenced by the hypothalamus, a part of the brain which is also involved in the regulation of emotional behavior and appetite. The possible involvement of the hypothalamus in Huntington's disease may contribute to the observed irregularities of hormone production and ravenous appetite.

The intriguing findings of endocrine gland changes in Huntington's disease and other neurodegenerative disorders demand further study and research support.

Genetics and Cell Biology

Looking at the brain with the aid of the neuroscientist's tools is obviously necessary in studying Huntington's disease and other neurodegenerative disorders. Still the questions remain: What causes the brain changes? Why do certain specific brain cells die at a particular time? Are the cell losses or the chemical changes the primary event caused by the Huntington's disease gene or are they a secondary process in reaction to some other mysterious change in the body?

The fact that Huntington's disease is a hereditary disorder provides a special advantage in answering these questions. Scientists know that whatever causes the myriad symptoms of the disease derives from one malfunctioning gene. Further, they know that genes control the manufacture of vital body chemicals—enzymes, proteins, or protein products—or they are involved in regulating other genes which do.

Although the major findings in Huntington's disease involve changes in the brain, there is reason to believe that other organs are involved. Many genetic disorders involve subtle changes throughout the body. Moreover, Huntington's disease is a dominant disorder, and many geneticists believe that such disorders involve abnormalities in the proteins which determine the structure of tissues found throughout the body. These include the proteins found in membranes surrounding the cell.

The search for a defective gene product, such as an abnormal membrane protein, can make use of highly sophisticated techniques which can distinguish among thousands of different proteins by virtue of their electrical and physical properties when exposed to an electric field.

It is the only hope they have — research— to lift this black cloud hanging over their heads and give them hope.

This is my prayer.

Other attempts to detect the abnormal gene product in Huntington's disease have involved comparisons between skin cells, red blood cells, or platelets (an element in blood) derived from Huntington's disease patients and normal controls. Some intriguing findings have been reported. (See Vol. II.) In addition to aiding the understanding of the basic disease process in Huntington's disease, research on tissues outside the brain can be of great value as a diagnostic test for the disease. Diagnosis now is based on observing choreic movements in someone with a family history of the disorder. A test which revealed an abnormality specific for Huntington's disease in easily accessible tissue, such as blood or skin cells, might also be a means of detecting carriers of the Huntington's disease gene. (See p. 24 for discussion of a presymptomatic test.)

Support of research to disclose abnormalities in tissues outside the brain is urgently needed to elucidate the mechanisms of Huntington's disease and related brain disorders.

Immunological Factors

Two immunological approaches have been explored in studying Huntington's disease. They are based on the hypothesis that the disease may involve an inappropriate response of the body's immunological defense system. Cells which normally attack invading organisms or foreign tissue are turned against the body's own tissue or do not provide proper immunological protection. *Further studies of immunological anomalies in Huntington's disease patients merit exploration.*

Virology

The discovery that a virus could give rise to chronic and fatal neurodegenerative disease startled investigators a few years ago. The Nobel laureate, Carleton Gajdusek, and others established that scrapie in sheep, a condition affecting mink, and two human disorders—kuru and Creuzfeldt-Jakob disease—make up a group of subacute or "slow virus encephalopathies." The possibility exists that Huntington's disease might also be a member of this group. Even though there are major differences between the two diseases, Huntington's disease shares with Creuzfeldt-Jakob disease the characteristics of late onset, dementia, and abnormal movements.

In preliminary studies Huntington's disease brain material has already been injected into animals, particularly the primates, to search for a possible transmissible agent.

Studies supported by the NINCDS and currently underway to search for a viral cause of Huntington's disease should be expanded and continued until a definitive answer is achieved.

Animal Models

No satisfactory animal model, either present in nature or artificially induced, has as yet been demonstrated for Huntington's disease or for most neurodegenerative disorders. The creation or identification of such a model would be of immense importance since it would permit a wider range of experimentation than is possible with human patients.

Several experiments offer promise in developing models for some aspects of Huntington's disease. One intriguing experiment reported recently involves injecting kainic acid in rats and monkeys. This acid destroys the same cells in the animal brains as are destroyed in Huntington's disease. Although the behavioral and motor symptoms in rats and monkeys are unlike the human symptoms and the condition is not hereditary, the kainic acid model may be the first biochemical model of certain features of Huntington's disease to be produced in other species. *Continued study of this model is recommended along with the search for other animal models. Typical of this is the study of certain strains of mice developed at Jackson Memorial Laboratories in Bar Harbor, Maine, and of Kerry Blue terriers with hereditary ataxia which are being studied at the Harvard Medical School.*

In summary, the Commission recommends that basic research in the following areas be supported:

- *Research should be expanded to delineate the neurochemical, structural, and functional organization of those brain regions (particularly the basal ganglia and cerebral cortex) affected in Huntington's disease.*

- *Neurochemical and structural pathologic analyses should be conducted on at-risk individuals and on Huntington's disease patients in the early stages of the disease in order to establish cause-and-effect relationships.*

- *Efforts should be made to perfect more sensitive electrophysiologic tests of basal ganglia functions which can be used in animal studies and in a noninvasive manner to monitor individuals having, or suspected of having, Huntington's disease.*

- *Drugs should be developed that are capable of selective enhancement or antagonism of those neurotransmitter systems associated with the motor and behavioral symptoms of Huntington's disease.*

- *Research should be supported concerning the response of the basal ganglia to drugs. Treatment criteria and protocols should be established for a nationwide collaborative study of potentially beneficial therapies for the treatment of Huntington's disease. Detailed evaluative guidelines for long-term studies should be developed to confirm or deny the value of each drug in the alleviation of neurologic and behavioral symptoms of Huntington's disease.*

- *Endocrinological profiles of Huntington's disease patients should be undertaken to determine whether there are sufficient endocrinological abnormalities to warrant additional investigation of the endocrine system as an index to the pathogenesis of the disease.*

- *Increased research is needed on the biology of cell growth and membrane kinetics, particularly as seen in the phenomenon of contact inhibition in Huntington's disease and related disorders.*

- *Support of tissue culture studies is urged to determine the nutritional requirements of Huntington's disease cells.*

- *Research is strongly recommended to discover if there are any electrophoretically detectable abnormal membrane proteins*

or other abnormal proteins in Huntington's disease patients.

- *Studies currently underway to search for a viral cause of Huntington's disease should be expanded and continued until a definitive answer is achieved.*

- *Serum antineuronal antibody and lymphocyte sensitization experiments in Huntington's disease patients should be continued and research to explore other immunological involvements in the origin or progression of Huntington's disease should be supported.*

- *Research should be supported to identify naturally occurring animal models of Huntington's disease as well as to experimentally create such models.*

My greatest concern is that real effort be made to encourage research to find some way of halting or making less difficult this disease. I realize that much research is now going on and only pray that more resources may be directed into this effort. It may be too late to help my son, but perhaps there is hope for future generations.

It is important to support basic research into descriptions of neuorotransmitters and their cellular interactions, particularly of peptide and other less well described transmitters. We need to develop the technology to identify these transmitters and their receptors in various areas of the brain, as has been done for the catecholamines and acetylcholine. We need to understand more about the cause and progress of senescence in nerve cell populations.

If we had enough money to give to research maybe they can find a cure for it before we are old enough to get it. I feel I have no time to worry, just to help raise money for the people who have it. Everybody can help if they try.

Clinical Research

Human research subjects are essential in advancing understanding of a uniquely human disease. As there are no true animal models of Huntington's disease, important research in understanding the disease must be conducted on human volunteers. Research patients must be studied by clinical investigators to learn about such complex and critical questions as what physiological functions may be altered by the disease, what hormones may be affected, how diet and environmental factors affect the disease, how thinking and feeling are affected by biochemical processes, how individuals cope with chronic degenerative disease, and many other challenging questions.

Clinical research patients are essential in efforts to find effective treatment. Drug studies must ultimately be conducted on human beings to determine the efficacy and safety of new medications and the nature and importance of side effects. Much of this research will require long-term study and comparison with patients with other neurodegenerative diseases or with normal controls. *As a vital part of research to understand and treat human brain disease, support for clinical research must be increased.*

New Technology

New technologies for diagnosis of disease and better understanding of brain functions in the living patient have created major scientific breakthroughs—but they require the utilization of human subjects. A major advance in the last decade has been the development of a new X-ray technique for studying the brain—computerized axial tomography (CAT). Using this technique investigators can identify a brain tumor, a blood clot or hemorrhage, and the loss of cells as seen in Huntington's disease. Because the X-ray exposure is very low, the CAT scan can safely be used repeatedly to trace changes in the brain throughout the course of the disease.

CAT scans are currently aiding in the diagnosis of Huntington's disease. Preliminary investigations are now underway to determine how early in the course of the disease the CAT scan can be of use. These studies and others utilizing computerized tomography must be carefully evaluated before they are given wide use.

"Computerized positron tomography" represents the apex of sophistication in this technique. When minute amounts of radioactive isotopes are injected or inhaled, positron tomography can reveal actual physiologic processes taking place in the deepest regions of the brain. This method may be effective in suggesting new biochemical, structural, and functional leads for understanding the origin, course, and possible treatment of many brain disorders, as well as greatly clarifying clinical findings. *Research to explore the design and distribution of equipment, methods of operation, analysis of data, and applications of computerized positron tomography should be supported.*

Genetic Linkage Studies

Studies of large multigenerational families with Huntington's disease are the only method now available by which to attempt to identify on which chromosome the Huntington's disease gene lies and to locate its exact position.

Classical family genetic linkage studies can progress immediately since they do not depend on biochemical understanding of the genetic defect. Successful linkage analysis could also determine whether there are multiple but very similar genes which produce different forms of Huntington's disease. If the location of the Huntington's disease gene on a chromosome were to be found, it could lead to the development of an animal model. *Classical genetic linkage studies should be supported.*

Behavioral Studies

The emotional and intellectual aspects of Huntington's disease are of great clinical and scientific significance. They bear on problems of diagnosis, early detection, treatment, and research on psychiatric and neurologic illness, as well as on the evaluation of pharmacologic agents. Only recently have the dementia and emotional disturbance associated with Huntington's disease received increased scientific attention, although they are the most distressing symptoms to the patient, the at-risk individual, and the family.

Research to develop psychological tests to correlate behavioral changes with neurophysiological and neurochemical abnormalities

in Huntington's disease must be encouraged and supported. Equally important is the need to learn what enables some patients, families, and individuals at risk to cope successfully with the emotional strains produced by Huntington's disease and other dominant genetic disorders, while other individuals, even those who never develop the disease, are psychologically shattered. Can good coping strategies be taught to affected families?

Epidemiological Studies

No reliable data exist on the overall incidence and prevalence of Huntington's disease. The few classical genetic and epidemiological studies that have been done were done many years ago before Huntington's disease came to public notice. They have only confirmed the view that widespread misdiagnosis, inaccurate and poor reporting, and even concealment of the disease make it presently impossible to know how many individuals have, or are at risk for, the disease. Leading epidemiologists who comprised the Commission's work group on epidemiology and biostatistics confirmed this systematic underreporting.

The number of families in Kansas affected by Huntington's disease is much larger than our statistics from death certificates would indicate.

Public testimony and anecdotal evidence gave abundant examples. For instance, while a state-by-state analysis of national mortality figures showed no reported deaths from Huntington's disease in one state over a period of years, the Huntington's disease health voluntary organizations had assisted families in arranging autopsies in that very state. These autopsies verified Huntington's disease as the cause of death.

A large-scale and detailed study on the prevalence and incidence of Huntington's disease in the United States is needed. Much information could also be gathered from a follow-up of earlier epidemiological and genetic studies of Huntington's disease in Minnesota (1951) and Michigan (1958).

Health Services Delivery Research

As new techniques and equipment have developed in the laboratory, scientific investigators and practitioners have become increasingly aware of delays or complications in applying them to patients in need. This includes not only concern for lags between the completion of research and general availability of innovations, but also the lags between the first clinical application of a discovery and widespread use by practicing physicians and acceptance by patients. The transfer of technology has become a matter of concern at the NIH, within the Public Health Service (PHS), and to Congress and the general public. Research on the delivery of health services is gaining prominence as these services grow.

The Commission strongly favors research on health services and on their delivery, and urges increased support for this work. Two areas in particular should be investigated:

- *Methods of treatment and care of Huntington's disease patients and others with chronic neurological disorders*, particularly those who are psychologically impaired. Research should be conducted on innovative methods of treatment and care, including prosthetic aids, rehabilitation techniques, and the use of architectural design to provide protection and orientation. Guides should then be developed for nursing staff and other health professionals.

  Research should be supported on the development of training programs to improve self-help skills for patients and to guide family members in more efficient and effective methods of care. Families are, by choice or by default, the main providers of long-term care and their vital role should be acknowledged.

  Research should also be conducted on the optimal size and groupings of patients with different but related disorders in therapeutic groups and care facilities.

- *Health Services Delivery and Manpower.* The intramural program of the NINCDS should initiate and support research on the types of health services which are necessary for Huntington's disease patients and others with related disorders. Research should be conducted on optimal combinations of health care providers (medical and mental health specialists, physician assistants and nurse practitioners, rehabilitation and speech therapists, and others) and on how services should be delivered.

The NINCDS should utilize the facilities of the NIH Clinical Center, the new Ambulatory Care Research Facility, and any other facility outside of the Government that it deems appropriate to conduct this research. (For further studies in health services, see Genetic Counseling, p. 77.)

The Commission recommends that clinical research in the following areas be supported:

- *New techniques now available should be employed and carefully evaluated in the study of patients with Huntington's disease and related disorders. Further studies to advance new technologies, particularly computerized tomography, and studies of the most practical, efficient, and cost-effective utilization of these technologies to accompany their development should be undertaken.*

- *Classical genetic linkage studies on Huntington's disease should be supported. International cooperation should be facilitated to obtain data and specimens from large family groups having inherited Huntington's disease from the same ancestor.*

- *Psychological test batteries should be developed to establish meaningful correlations between the arrays of neurological, neurochemical, and neurophysiologic abnormalities in Huntington's disease.*

- *Psychological tests and clinical studies, particularly longitudinal and/or cross-sequential, should be undertaken to define the development of abnormal behavior and coping behavior of Huntington's disease patients, those at risk, and their families.*

- *Behavioral studies of Huntington's disease patients and families should be included in existing centers for neuropsychological research and in the Huntington's disease and related disorders "Centers Without Walls."* (See p. 45.) *Frequent communication and interchange among investigators should occur so that data can be pooled, representative samples developed, and interdisciplinary teams formed without duplication.*

- *A large-scale and detailed study on the prevalence and incidence of Huntington's disease in the United States should be undertaken. Much information could also be gathered from a follow-up of earlier epidemiological and genetic studies of Huntington's disease in Minnesota (1951) and Michigan (1958).*

- *Research on health services delivery should be supported including research on methods of treatment and care of Huntington's disease patients and others with chronic neurological disorders, and studies of health services delivery and manpower. The intramural program of the NINCDS should initiate this research. Bed space in the NIH clinical facilities should be allocated flexibly according to need rather than on the basis of strict quotas among institutes.*

In addition to the biochemical and other types of research aimed at elucidating the basic biochemical error in Huntington's disease so that a pharmacological or other therapy can be devised, we need, on a more immediate basis, training of people in the provision of services to assist families in coping with the day-to-day problems of having the illness in the family.

Presymptomatic Detection Tests

No X-ray, EEG, blood sample, or any other biochemical technique nor any psychological test can distinguish carriers from noncarriers of the Huntington's disease gene. Prenatal diagnosis is not possible. Tests which have been tried—but which are either ineffective or too preliminary—include electroencephalographic studies, tests of psychological and behavioral abnormalities, tests measuring chemicals in cerebrospinal fluid, and genetic linkage studies.

Some recent experimental tests for presymptomatic detection involved giving L-dopa to individuals at risk for Huntington's disease. In one study of 30 subjects at risk, one-third developed abnormal chorea-like movements while none of the 25 normal controls were affected by the drug. *The Commission does not recommend this test because of a number of serious disadvantages:*

- A negative result does *not* rule out the presence of the Huntington's disease gene.
- It is not known whether its administration may actually precipitate the onset or accelerate the course of the disease.
- If symptoms are induced, serious psychological problems may result, possibly leading to suicide.

Above all, it must be emphasized that the "L-dopa test" was developed for experimental investigations only and was to be utilized solely by trained researchers. The Commission was disturbed to learn through public testimony that the test is being administered by some clinical practitioners, who provide no preparation or follow-up of at-risk individuals and who appear to treat the test as definitive.

A second more promising approach is now being investigated. Several researchers have found that GABA levels in cerebral spinal fluid of individuals at risk for Huntington's disease divide those individuals into two different groups. This follows the 50-50 expectation for at-risk people.

Further studies and long-term follow-up will be necessary to verify the validity of this and any other putative presymptomatic test. The accuracy of any such test is of utmost concern, particularly in the absence of adequate treatment.

Ethical Guidelines

The ideal test for presymptomatic detection would be a safe noninvasive laboratory procedure which would disclose the presence or absence of a specific protein or enzyme reflecting the genetic defect in Huntington's disease.

While urging that research to develop a presymptomatic detection test be given the highest priority, the Commission recognizes that advances in detection, understanding, and treatment may not occur concurrently. During the course of research in developing the test, patients who volunteer may be eager for information. *They may deliberately or inadvertently receive information about preliminary findings which, because they are unverified, could be extremely harmful.* To ensure that proper precautions are taken in the *research to develop a presymptomatic test,* the Commission has established a set of guidelines concerning the conduct of research, instructions to volunteers, and the handling of preliminary information. The Commission urges their adoption by all investigators involved in research which could lead to a presymptomatic test.

If a presymptomatic test is developed and validated in the absence of effective treatment for Huntington's disease, the test results have the potential for disastrous consequences as well as good. It is this possibility that has prompted the Commission to develop *guidelines for the use of a presymptomatic test* once it is validated. (See Vol. II for guidelines.)

The Issue for All Genetic Diseases

The ethical issues raised by the development of a predictive test for Huntington's disease are instructive for all genetic health problems. New discoveries and techniques will increase the possibility of early diagnosis and screening programs, perhaps for diseases not even now regarded as genetic. Because medicine aims to prevent as well as to cure, early detection techniques will be used increasingly. There will not always be effective therapy at hand. A thorough examination of the problems raised by a predictive test for Huntington's disease will help focus the issues

and provide guidance for the resolution of the dilemmas relevant to many, particularly dominant, hereditary conditions.

The Commission unanimously takes the position that any presymptomatic testing or diagnosis must be entirely voluntary. No governmental agency or progam should require individuals to undergo such testing under any circumstances, nor as a condition of receiving any benefit.

The Commission recommends that in view of the immense social and scientific urgency in developing a dependable, objective, and reproducible laboratory test to differentiate between those at-risk individuals who do and those who do not carry the gene for Huntington's disease, it is essential that each promising lead be explored and exploited by competent scientists.

Being at risk is never knowing. Even though I'm statistically young, when I get nervous about anything or uptight, the tendency to think that it is H.D. is awfully strong . . . I don't think I could ever emphasize the importance of some form of therapy to be made available to cover these particular areas . . . I want help to understand and to cope without messing up my life any more than it is.

My sister and her husband had four children, ranging in age from approximately 21 to 12, when it was discovered that he had H.D. The oldest girl completely separated herself from the family, married and had a baby, eventually committed herself to a mental institution and deserted her son (in the light of the fact that she herself is now a victim of H.D., this can be more easily understood); the second girl suffered a nervous breakdown; the oldest son withdrew and became involved with drugs for awhile; the youngest son seemed to suffer the least damage to his psyche at that point.

One of the hardest things for me to do is to look at my three darling girls and to think what I may have given them. If we had only known, we would have adopted children. I love my girls more than life but I cannot stand the thought of seeing them rot away with H.D. If you want to know what hell is like, it's seeing someone you love with H.D.

We don't tell anyone what my husband had. We feel if we do, the boys would be watched for the same telltale signs of geting the disease. I want them to lead a normal life as long as possible.

I used to dream that I was being forced to choose which one of my children would have Huntington's disease. I don't do that anymore, thank heavens.

Huntington's disease can ruin your life. It makes you feel as though there is no future and no hope for you or your family. You see yourself as your parent is now.

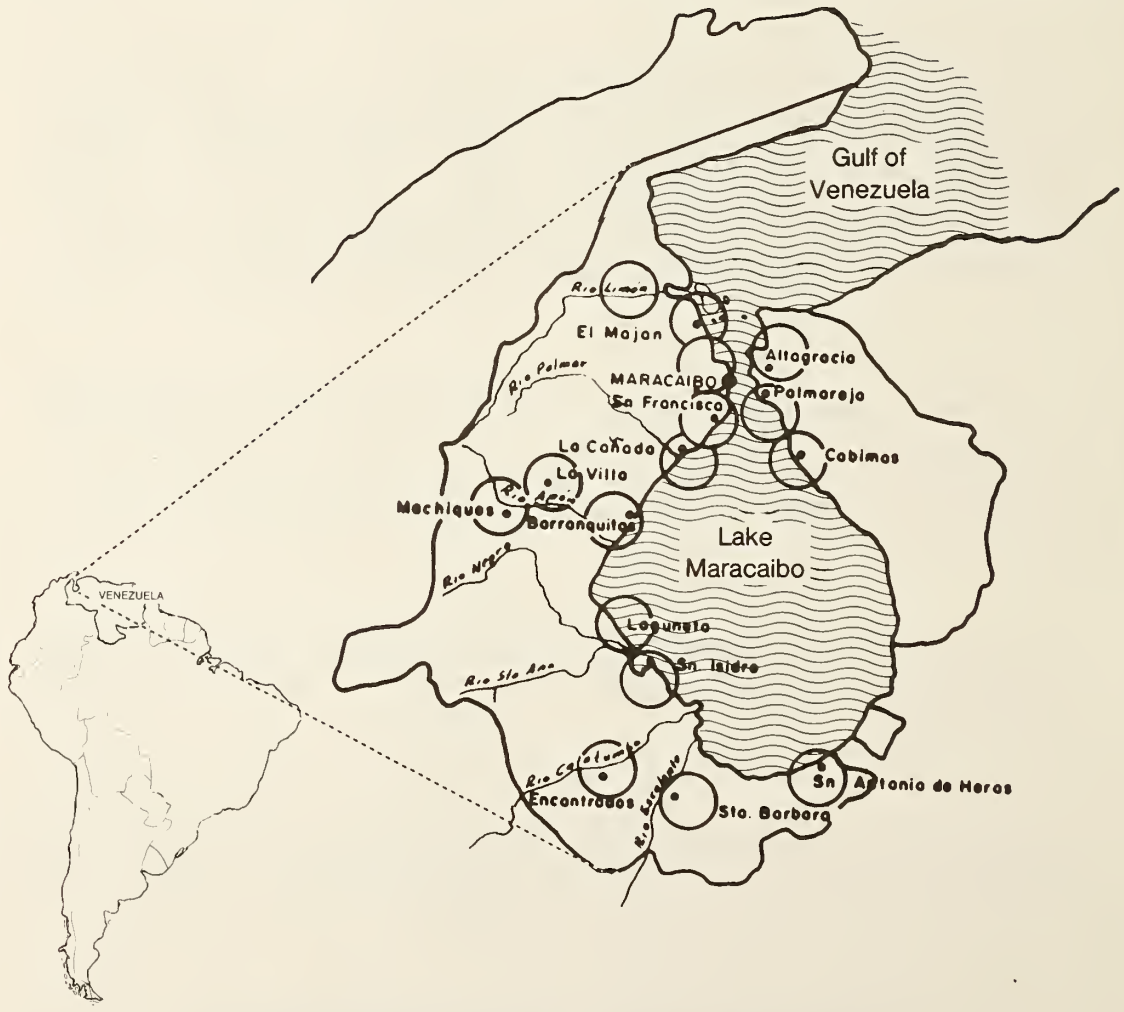
Special Research Activities

The Commission found the following research and research-related activities particularly worthy of support.

The Venezuela Project

Basic and clinical research on the genetic defect in hereditary disease can be greatly facilitated by the study of isolated communities where families with dominant genetic disorders have intermarried. Focusing on Huntington's disease, if both parents are patients, there is a chance that some children born of the marriage may inherit the defective gene from both parents. If such a child survives, the "double dose" of the gene might well trigger an earlier and more dramatic onset of the disease. Further, since there would be no normal gene to counteract the effects of the deleterious gene, whatever goes wrong in the body's cells might be more readily revealed in chemical and other analyses of body tissue or fluids.

This reasoning accounts for one of the few recommendations the Commission is making that applies exclusively to Huntington's dis-



Communities of Choreics in the State of Zulia

ease. For there is just such a group of Huntington's disease patients who have frequently intermarried living in communities near Lake Maracaibo in Venezuela. All patients there are descended from a single individual who introduced the disease into the area in the 1860s. These patients provide a unique and invaluable asset for research in neurochemistry, pharmacology, cell biology, genetics, genetic linkage, and other major disciplines. The group should also be studied by behavioral scientists and ethnographers to explore their beliefs, attitudes, customs, and ways of coping with the disease. They are a poor and socially ostracized group who would benefit from the attention and care provided by specialists in Huntington's disease.

The Commission recommends that the Congress appropriate funds to the NINCDS to enable that agency to collaborate with Venezuelan scientists to design, support, and conduct an interdisciplinary study of the population affected by Huntington's disease in the state of Zulia, Venezuela.

Patient Rosters

An indispensable aid in research are carefully maintained, accurate, and up-to-date records of patients who have indicated their willingness to participate in research projects. The private health voluntary organizations for Huntington's disease have acted as informal intermediaries between their members and clinical scientists. These organizations maintain partial lists which are less useful than they could be because the voluntary organizations do not have the resources to organize the available data.

The Commission recommends that listings of patients and their families be maintained on a strictly voluntary basis as the responsibility of a state or a private health agency (health voluntary organization or medical school). Support to ensure accurate and up-to-date records should be provided by contract with the NINCDS. To ensure protection for individual privacy rights, the general provisions contained in the recent National Bureau of Standards Special Publication, A Policy Analysis of Citizen Rights Issues in Health Data Systems, *should be observed in establishing and maintaining listings of patients and families.*

National Tissue Bank

Quick and easy access to carefully collected, stored, and documented tissues and biological fluids taken at autopsy or from the living patient is critical in performing research on a disease for which only human tissues will suffice. The most valuable and precious tissue needed in research on Huntington's disease and related disorders is brain tissue. Except in very rare circumstances, this can only be obtained at autopsy.

Cerebral spinal fluid (CSF), which bathes the brain and gives some information about its activities, is also critical for research.

Individual scientists usually do not have the time or the resources to collect autopsy material or spinal fluid for extensive experimentation. To obtain sufficient quantities of this scarce material, tissues must be collected by specially trained professionals who can maintain and distribute the samples according to precise protocols.

The recommendation for a National Tissue Bank was independently and strongly urged by every scientific work group established by the Commission and was repeatedly proposed in letters sent to the Commission by leading scientists throughout the world. The Tissue Bank would collect, preserve, and distribute brain and other central nervous system tissues and other body tissues and fluids from patients with Huntington's disease, from those with related neurological and psychiatric disorders, and from normal controls. These latter tissues are essential for purposes of comparison and in order to establish the specificity of findings. The Bank should serve investigators both throughout the country and internationally.

Although the purpose of the Bank would be to distribute tissues for scientific study, some tissues, such as blood serum or spinal fluid, could be stored so that tissues from many family members with hereditary disease could be accumulated over several generations. As new hypotheses are developed requiring confirmation by many generations of a single family, investigators can return to these samples.

The Huntington's disease health voluntary organizations have also unanimously supported the idea of a Bank and have already begun to educate their members regarding the legalities of tissue donation. Special organ donor cards have been developed and tissue donation teams organized locally to assist families.

Accordingly, the Commission makes the following recommendation:

The Commission recommends that Congress provide funds and direct the Secretary of the Department of Health, Education, and Welfare (DHEW) to establish a National Tissue Bank for the collection and distribution of tissues for research on Huntington's disease and related neurological and psychiatric disorders. (See Vol. II for detailed guidelines for the Bank.)

A small amount of money, or at least a modest amount, can catalyze resources into action which are already poised and just hampered because the necessary connecting links are not there.

Since most of the relevant research results in Huntington's disease have been found on post-mortem brain tissue it would seem wise to pursue the development of a brain bank facility.

I believe that the highest priority should be given to obtaining first-rate central nervous system tissue specimens for state-of-the-art studies of neurochemistry and neuropathology.

Procedures for obtaining brain tissue from patients with Huntington's disease and other genetic diseases are rather haphazard and it would be excellent if a coordinated effort could be developed so that tissues were not lost for further research.

Exchange of Research Ideas

Interdisciplinary workshops, formal conferences, and symposia are different but important means of stimulating new ideas, bringing attention and focus to a problem, rapidly sharing new knowledge, and consolidating a state of the art.

Interdisciplinary Workshops

Sometimes the most modest suggestion for resolving a problem has the potential to produce the greatest reward. The recommendation for informal interdisciplinary workshops as a concept to stimulate fresh ideas in any field and recruit scientists for that field was recommended to the Commission by all its work groups, including those involved with social management as well as basic and clinical research. Many letters received from scientists throughout the world recommending promising areas of Huntington's disease research suggested interdisciplinary workshops as a means of generating and exploring new hypotheses.

How do we get young scientists in the scientific community interested in a relatively little known disease? I've been extraordinarily impressed by the success of the workshops. Typically, when the young scientists come they are first shocked by the horror of the disease, but I think more importantly after several days of discussions, they realize that it's possible to do creative scientific work along their own lines of specialization on the problem of Huntington's disease.

Workshops are an effective and economic way of bringing challenging problems in science into sharp focus and catalyzing activity to solve them. They are a particularly potent stimulus to research in a disease like Huntington's disease, for the disease itself could be described as interdisciplinary: symptoms and findings cut across the major fields of genetics and neuroscience but have yet to yield a unifying hypothesis.

The experience of one of the private foundations in Huntington's disease in organizing and conducting workshops has been eminently successful and has demonstrated the value of the workshop concept. (See letters from participants in Vol. III.)

The judicious selection of themes of high interest and the choice of an enthusiastic combination of new and more experienced research and clinical scientists to discuss them have paid high dividends. Workshops have led to productive research projects, generated new and interesting hypotheses concerning the genetic defect in Huntington's disease, and stimulated young scientists to turn to neurodegenerative disease research.

I started out in 1975 on the workshop programs with H.D. and became utterly fascinated by the research potential that was there.

There are certain basic principles involved in creating a successful workshop. A research workshop involves bringing together an interdisciplinary group of investigators in an informal and open atmosphere for several days. This encourages a broadening perspective through the rapid exchange of information and ideas, stimulates the kind of free-wheeling speculation and hypothesis-building that are precluded at larger, more formal scientific meetings, and often inspires a young scientist who has not yet made a career choice to make a commitment to disease-oriented research. A skillfully organized workshop can accomplish that rare interchange among those with great medical knowledge, those with basic science expertise, and those involved in urgently needed high technology. This triumvirate of specialized skills is desperately needed in the complex world of disease research, where sadly, basic and clinical researchers often do not see their relevance for each other.

The proven success of the workshop concept in generating research hypotheses and attracting new and experienced investigators to assume the challenge of research in Huntington's disease prompts the Commission to recommend a program of interdisciplinary workshops in its National Plan. Workshops are flexible and economical. They can focus on problems in treatment and care of patients as well as basic research, and can easily apply to a variety of related genetic or neurodegenerative disorders in addition to Huntington's disease.

The Commission recommends that the NINCDS provide financial support, through its grant or contract mechanisms, for workshops conducted by an appropriate Government or voluntary agency to bring together an interdisciplinary group of basic scientists and clinicians to explore promising leads in research, care, and treatment of Huntington's disease and related disorders.

Neurogenetic Newsletter

Research scientists and clinical investigators studying Huntington's disease and related disorders have expressed the need for an informal publication to promote a more rapid and direct exchange of ideas than is currently available through journals or Government publications of a more general nature. The newsletter would include information on current research on Huntington's disease and related disorders (publicly and privately sponsored), lists of active investigators, negative findings, brief communications, and other relevant information. It would serve to facilitate research and stimulate communication among individuals primarily interested in the genetic neurodegenerative disorders.

The Commission recommends that the NINCDS sponsor, through its grant or contract mechanisms, a publication designed to provide current brief bulletins on research, management, and related information for professionals interested in Huntington's disease and other genetic or neurodegenerative disorders.

Conferences and Symposia

Conferences and symposia are of prime importance in generating interest in a research field. The Centennial Conference on Huntington's Disease held in Columbus, Ohio, in 1972 stimulated research nationwide. Proceedings of the conference were purchased by scientists and libraries at a rate far exceeding expectations.

Five years have passed since that major conference. Ideas have been verified that were only speculative at the time. The momentum generated by the Ohio Conference has produced excellent research. In order to provide a forum to report these developments and discuss common interests, the Commission believes that the time is now right for a second international symposium on the research and management of Huntington's disease.

The Commission recommends that the NINCDS sponsor an international symposium on Huntington's disease to take place within the next fiscal year (FY 1979). The Commission further recommends that such symposia take place every five years following as long as such forums prove useful.

Publications should be fostered. I worry that many findings important to our understanding of neuronal functioning are hidden away in notebooks because the data were never completed for formal publication. Possibly an informal bulletin could be put together on a monthly or quarterly basis encouraging researchers to share these bits and pieces of information. I would also sponsor a yearly book on topics in neurogenetics which would bring together important advances made in population genetics, animal models, drug studies, biochemical analyses, etc. which directly bear on our understanding of inherited diseases affecting the nervous system.

Resources for Research

Small Grants Program

A "small grants" program should be established in the NINCDS for the purpose of supporting exploratory research in the neurosciences. These awards would allow scientists to pursue unconventional new approaches, pilot-test a hypothesis, exploit promising applications of developing technologies, or enter a new field after working in one area for some time. Under existing programs such fresh starts or new leads are difficult to fund. The "small grants" program would provide awards up to $10,000 in direct costs and reasonable indirect costs to investigators. This would permit research for a limited period to determine the feasibility of a full-scale project or to assess the validity of a new hypothesis. Funds from these awards might be used to enable investigators to spend some time in another's laboratory or work with someone to learn new techniques or applications of equipment.

Seed money serves to stimulate new approaches to problems. And support for young investigators serves two purposes. It directs research to a particular problem, of course, but it also focuses the attention of young persons on the problems of genetic disease at a formative stage in their scientific development, and extracts a commitment from them in their further work.

Applications would be subject to strict peer review by an initial review committee and would be approved by the NINCDS Advisory Committee. (See Vol. II for criteria for selection and review.)

The Commission recommends that Congress establish a "small grants" program in the NINCDS to foster pilot or small-scale projects to explore new scientific technologies or test the feasibility of innovative research hypotheses in the neurosciences in general.

General Clinical Research Centers

The Division of Research Resources (DRR) of the NIH operates the General Clinical Research Centers Program (GCRC). The Program was established in 1960 in response to the need for specialized facilities and trained personnel required for high quality clinical research. Funds for the GCRC Program are intended primarily to establish and maintain, mostly in medical teaching institutions, separate discrete hospital units in which clinical research can be pursued. Center grants support an average of 10 patient beds per unit and ancillary laboratory, dietary, and personnel support facilities. Provision for outpatient research is now being included. Patients who are admitted to the Center for study purposes only pay no charges. Salaries for some professional staff are included in the GCRC grant. Investigators conducting research projects in the GCRCs are supported by NIH or other funds.

The GCRC Program provides a unique setting for investigators from many different disciplines to study a variety of disease problems using common resources. The program centralizes resources, encourages interdisciplinary cooperation, and is cost effective. Centers have enabled investigators to gain insights into disease processes and speed the application of research findings to patient treatment and care. They have also served as a training resource where medical and paramedical students can gain essential experience. Indeed, in the recommendation for a unique pilot program, "Centers Without Walls" (see p. 45), the Commission suggests that an existing GCRC might be used as the core of a clinical facility for research on Huntington's disease and related disorders.

Much of the invaluable clinical research on Huntington's disease would not have been possible without the GCRC Program. Investigator grants usually do not pay for patient costs and public or private insurance plans do not cover hospitalization for research purposes.

Funding for the GCRC Program is in serious straits, however, having been held to a constant level of $42 million since 1972. As a result, the number of research beds available has decreased by 14 percent from 907 in 1972 to 779 in 1976.

In 1976 only slightly more than $50,000 of GCRC funds was spent on Huntington's disease research. This amount must increase as more investigators become active in clinical research on Huntington's and related diseases.

In addition, the Commission believes that the GCRC Program is essential in maintaining high standards of clinical research in general in the United States.

One of the greatest weaknesses financially in research in this area is the absence of research beds. You can't put research bed costs on grants because the Government won't accept them. They just cut them out. You can't ask the hospitals to underwrite them—they simply can't afford it—and you can't get third party insurance, like Blue Cross, to pay patient costs if the patient's stay in the hospital is prolonged for the sake of research, even if the research is ethical and is ultimately designed to help that patient and others with the same disease. So nobody can pay for it and the research doesn't get done. I know that there are projects we would do in schizophrenia and in Huntington's disease if bed costs could be paid.

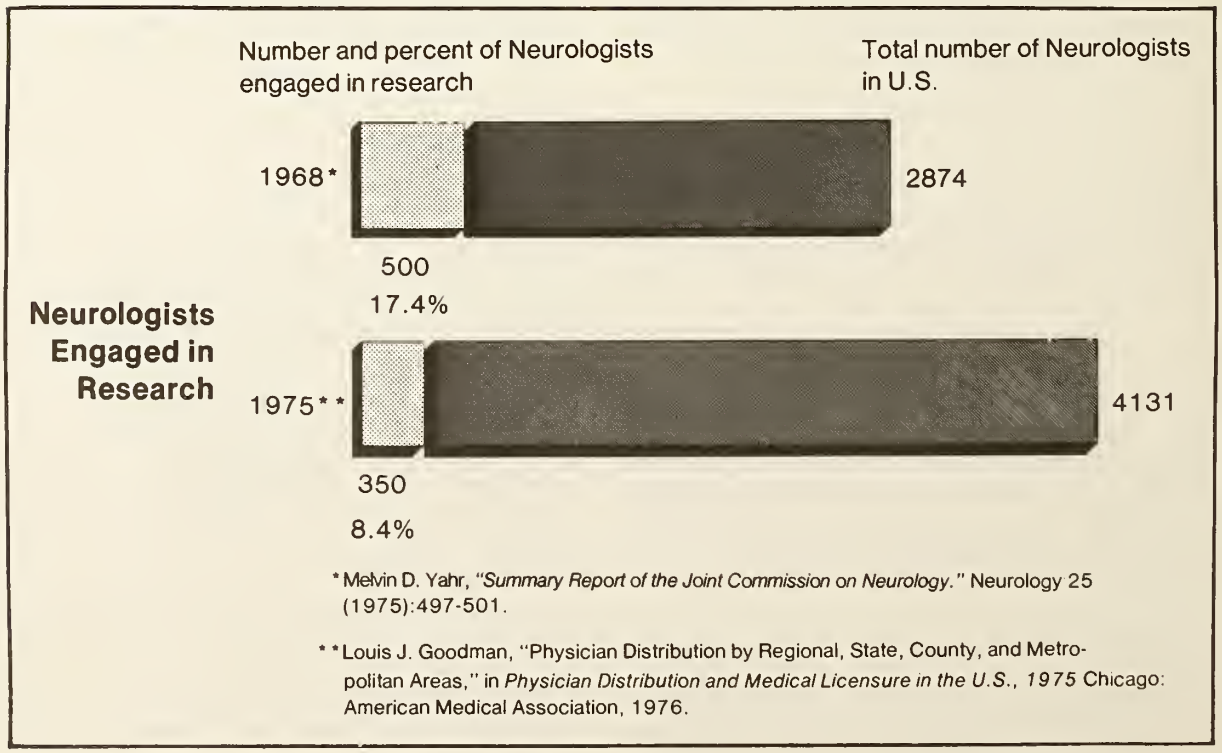
The Commission recommends that funding for the GCRC Program be commensurate with clinical research needs and take into account not only the rising costs of inflation but also the value this program offers as a service delivery mechanism.

Research Training

To maintain the high quality and productivity of research in the United States it is essential to replenish the pool of scientific manpower with innovative and well-trained young investigators. Unless there are well-designed and stably funded training programs offering incentives and opportunities, the best minds will not be attracted to research.

Unfortunately, support for training programs has dropped from a high of $169 million in FY 1969 to $122 million in FY 1976. The number of persons in training programs is down 25 percent from a high of 14,122 in 1969 to 10,546 in 1976. The decline in certain areas, such as clinical research, is particularly severe and raises the threat of critical manpower shortages ahead. The numbers of *physician-researchers* have diminished substantially. In 1971 there were 4,779 post-M.D. degree holders receiving NIH research training stipends and fellowships in clinical research. In 1975, four years later, the number had dropped to 2,797—a 40 percent decline—suggesting an acute shortage of clinical researchers in the future, with a resulting decrease in research on the development and application of new treatments.

The need for clinical investigators is particularly important for research on Huntington's disease and related neurological and genetic disorders for which there are no animal



*Melvin D. Yahr, "*Summary Report of the Joint Commission on Neurology.*" Neurology 25 (1975):497-501.

**Louis J. Goodman, "Physician Distribution by Regional, State, County, and Metropolitan Areas," in *Physician Distribution and Medical Licensure in the U.S., 1975* Chicago: American Medical Association, 1976.

models. Because research on Huntington's disease is a new and growing area there is a need to stimulate young investigators to attack the problems it and allied disorders present. Reductions in the research training budget and deterrents in current programs of training support, such as the inability of clinical research to serve as a payback for Federal medical school scholarships, dampen interest at a time when strong incentives are needed.

The best way to encourage the development of new scientific ideas is the provision of money to provide stable support for mature investigators and to provide funds for young men and women to be trained appropriately and to give promising new investigators a start. The past few years have been little short of disastrous and it is becoming increasingly difficult to attract bright young neurologists into research careers.

The NINCDS is the principal institute of the NIH for research on Huntington's disease and in the neurosciences. From 1969 to 1976 the total number of post-doctoral research trainees in NINCDS programs dropped by almost half—from 1,120 to 609—while the number of M.D. post-doctoral research trainees dropped by more than half—from 912 in 1969 to 454 in 1975. The Joint Commission on Neurology of the American Neurological Association and the American Academy of Neurology pointed out in their report, *Neurology Manpower—A Survey*, that there were fewer than 400 neurologists who reported research as their primary professional activity in 1971. Citing the American Medical Association, *Distribution of Physicians in the United States*, this represented a drop of over 100 (or 18 percent) from 1968, although the total number of active neurologists increased by 24 percent. The report states, "The decline in the absolute number of neurologists engaged in research represents an ominous development for advancing our fundamental information about the nervous system and even more for the clinical benefits to be derived."

The groundswell of interest in neurosciences research cannot be maintained without adequate research manpower. More critically for Huntington's disease, important new leads generated by the current surge of creativity in the neurosciences face the prospect of an insufficient number of new researchers to explore and exploit them. Ultimately the lack of adequate training support saps the vitality of all neuroscience research and forestalls the hope of treating or preventing major illnesses.

The Commission endorses the policy of Federal support for research training, but finds present levels of financial support, methods of determining "areas of need," and provisions in the current National Research Service Awards Programs (NRSA) to be highly unfavorable for new and rapidly developing areas of research which promise important advances. Appropriate changes must be made so that research training support is more sensitive and responsive to the targeting and development of new scientific leads.

One of the greatest limitations of the development of our research is the difficulty to find salaries or positions for young scientists and fellowships for exchange of scientists among laboratories.

The Commission recommends that to ensure a continuous supply of highly qualified and well-trained investigators it is essential that:

- *Annual levels of appropriations to the NINCDS for research training under the NRSA Program be set at $9.3 million in FY 1979, $11.7 million in FY 1980, and $12.9 million in FY 1981.*
- *Annual levels of appropriations to the NINCDS for Research Career Program Awards be set at $3.5 million in FY 1979, $4.0 million in FY 1980, and $4.6 million in FY 1981.*
- *Annual appropriations for the NIGMS for research training under NRSA (including Minority Access to Research Careers) be set at $46 million in FY 1979, $48.3 million in FY 1980, and $50.7 million in FY 1981.*
- *Annual appropriations for research training to the other institutes conducting research related to Huntington's disease (NIMH, NIA) be increased.*

Research Funding

Throughout the 11 hearings of the Commission's public testimony, the single most sustained plea was for increased research on Huntington's disease. Research was the recommendation of highest priority. The demand for increased *research* was also the overwhelming request in a survey of families with Huntington's disease to determine their need for *services. Only through biomedical research can the cycle of disease and death be stopped.*

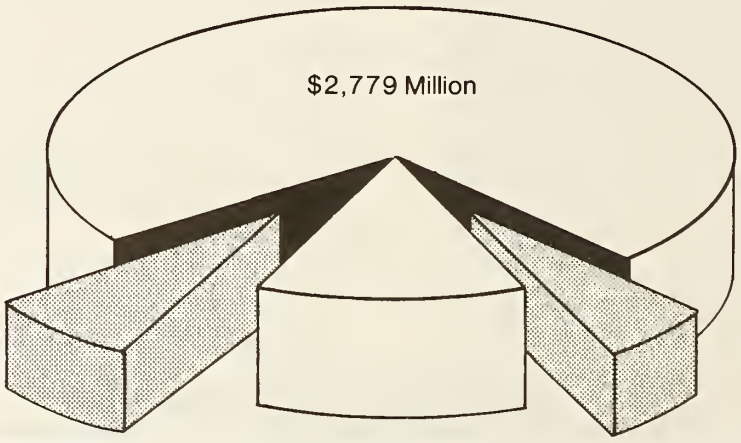
Yet just at a time when researchers have identified the neurosciences and genetics as the leading edge of biomedical research, and just when consumers are urging support for such research, there has been a major decline of funds for those institutes of the NIH most concerned with these fields:

- Funds appropriated to the NINCDS, the principal agency for nervous system research, are only about one-sixteenth of all NIH funds.
- Funds appropriated to the NIGMS, the institute mainly responsible for support of research on genetics, are only about one-tenth of all NIH funds.

Despite the recognized potential research in the neurosciences and genetics has for discoveries that could help millions of sufferers and save billions of dollars, appropriations have not even kept pace with inflation. As research dollars plummet, health care costs soar.

- Direct care costs for *mental illness* were *$17 billion* in 1975. Money used to investi-

**Federal Outlays for Health Research — 1976*
(In Millions of Dollars)**



Ranking of Diseases by Selected Incidence Measure, United States — 1971

Deaths	Days in Hospital	Incidents Causing Limitation of Activity
1. Heart and Lung	1. Arthritis, Metabolic & Digestive	1. Neurological & Visual
2. Cancer	2. Heart & Lung	2. Heart & Lung
3. Neurological & Visual	3. Mental Illness	3. Arthritis, Metabolic & Digestive
4. Allergy & Infection	4. Neurological & Visual	4. Mental Illness

*The Federal Health Dollar: 1969—1976, Center for Health Policy Studies, National Planning Association, February 1977.

**Chartbook of Federal Health Spending: 1969—1974, Center for Health Policy Studies, National Planning Association.

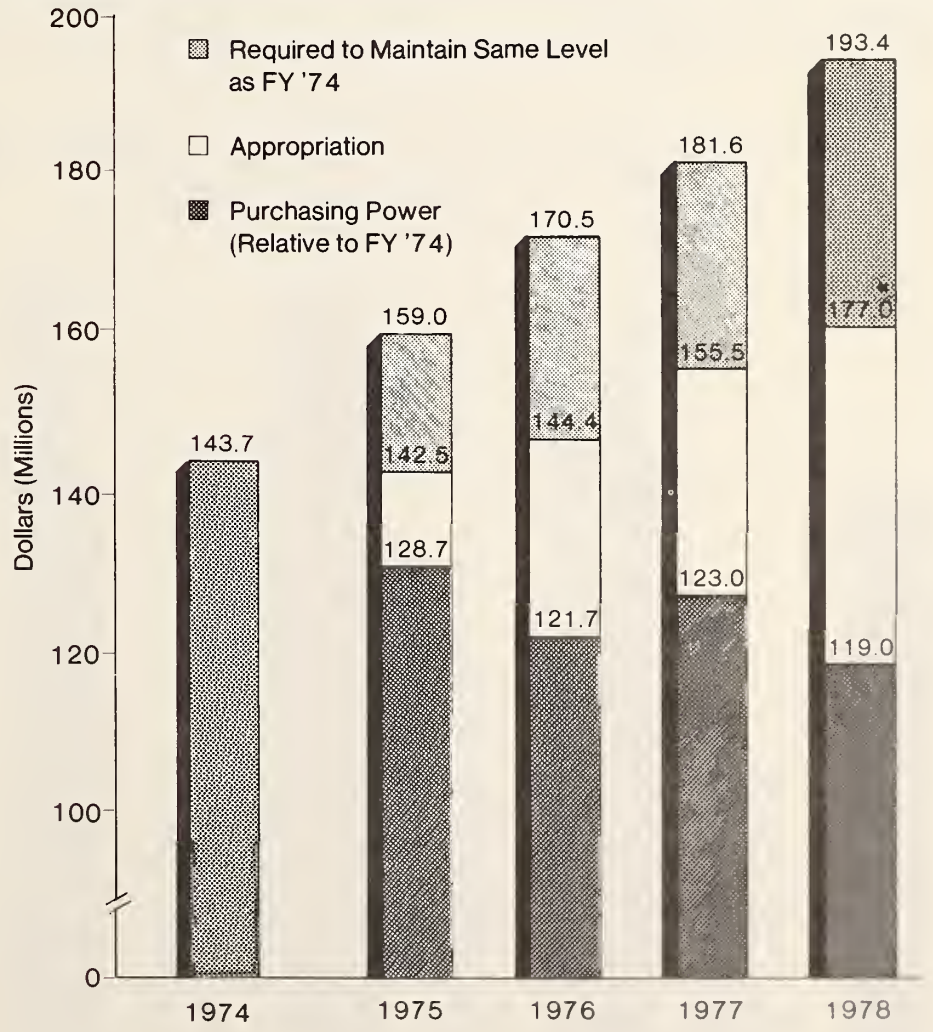
gate schizophrenia directly through NIMH has stabilized at approximately *$10 million* since 1969.

- Direct care costs for *genetic diseases* were estimated from *$3 to $5 billion* annually. Funds for research related to genetic disease total about *$100 million, or less than 5 percent of NIH research dollars.*
- More than *four million people* in the United States suffer from *serious* limitation of activity as a result of *neurological disorders.*
- *Neurological and visual impairments were* the *third leading cause of deaths* and the *leading cause of limitation of activity* in the United States in 1971.

The NINCDS and Huntington's Disease

Without adequate financial support for the scientific research outlined in this report, there is little possibility of achieving understanding, control, and, ultimately, elimination of Huntington's disease and other neurological and genetic disorders. Approximately $1.2 million of the NINCDS budget goes to research directly and indirectly related to Huntington's disease, which is less than 1 percent of the total NINCDS budget. NINCDS currently (FY 1977) is funding only three research projects on Huntington's disease. *Limitations on the NINCDS budget do not allow sufficient flexibility to explore new and promising leads in research.*

Effect of Inflation on NINCDS Budget



Source: Office of Program Planning and Evaluation, NINCDS.
*Information from Division of Financial Management, NIH.

The NINCDS Funding Dilemma

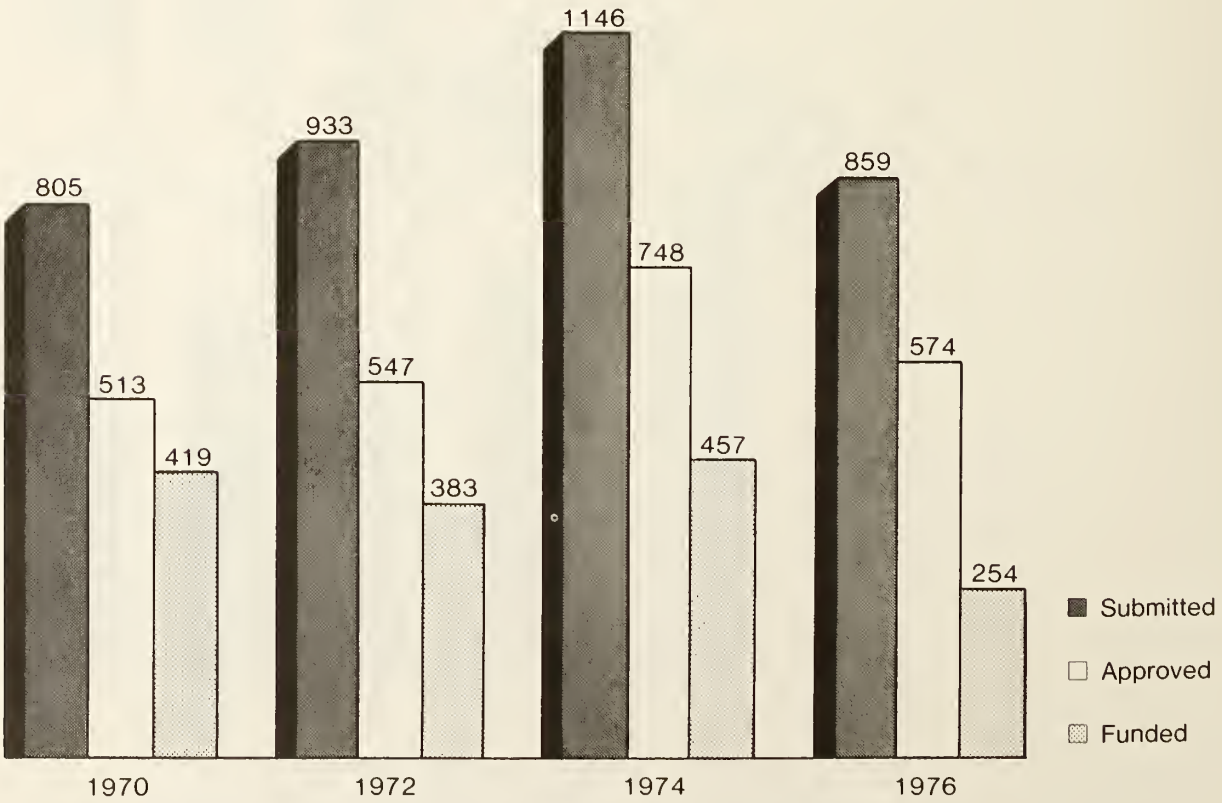
The NINCDS has primary responsibility for supporting research on a host of neurological disorders which cost approximately $70 billion annually (for all costs). In 1977, the Institute's budget was only $155.5 million. It was able to fund less than one-fourth of *approved* grants. Inflation has further eroded the institute's constricted funds, reducing the institute's actual purchasing power in constant dollars to less than it was in 1971. Not only does this precarious funding situation jeopardize good research in Huntington's disease, it also threatens research on Down's syndrome, senile dementia, multiple sclerosis, muscular dystrophy, Parkinsonism, cerebral palsy, amyotrophic lateral sclerosis, and many others. These are just a few of the disorders studied at the NINCDS, which is also the institute that conducts research on hearing, pain, sleep disorders, brain dysfunction and learning disabilities in children (dyslexia), stroke, epilepsy, headache, spinal cord injuries, and a variety of infectious diseases of the nervous system such as shingles and encephalitis.

The basic genetic fault of this disease is not known. Therefore, in addition to following the few biochemical leads now known, it is crucial to support basic research in the neurosciences, and in genetics.

For the same reason, it is crucial to support research related to other movement disorders, other forms of dementia, and the biologic basis of aging of the brain.

Other institutes of importance in research on the nervous system and genetics face comparable problems of inflation: the NIGMS (the lead institute for genetic studies), the NIMH, the NIA, the DRR of NIH, and the VA. They, too, must be supported at levels which will allow funding of excellent research projects.

Competing Research Grant Applications Submitted, Approved, and Funded by NINCDS FY 1970 — 1976*



*Source: Statistics and Analysis Branch, Division of Research Grants, National Institutes of Health.

Decline of the Neurosciences?

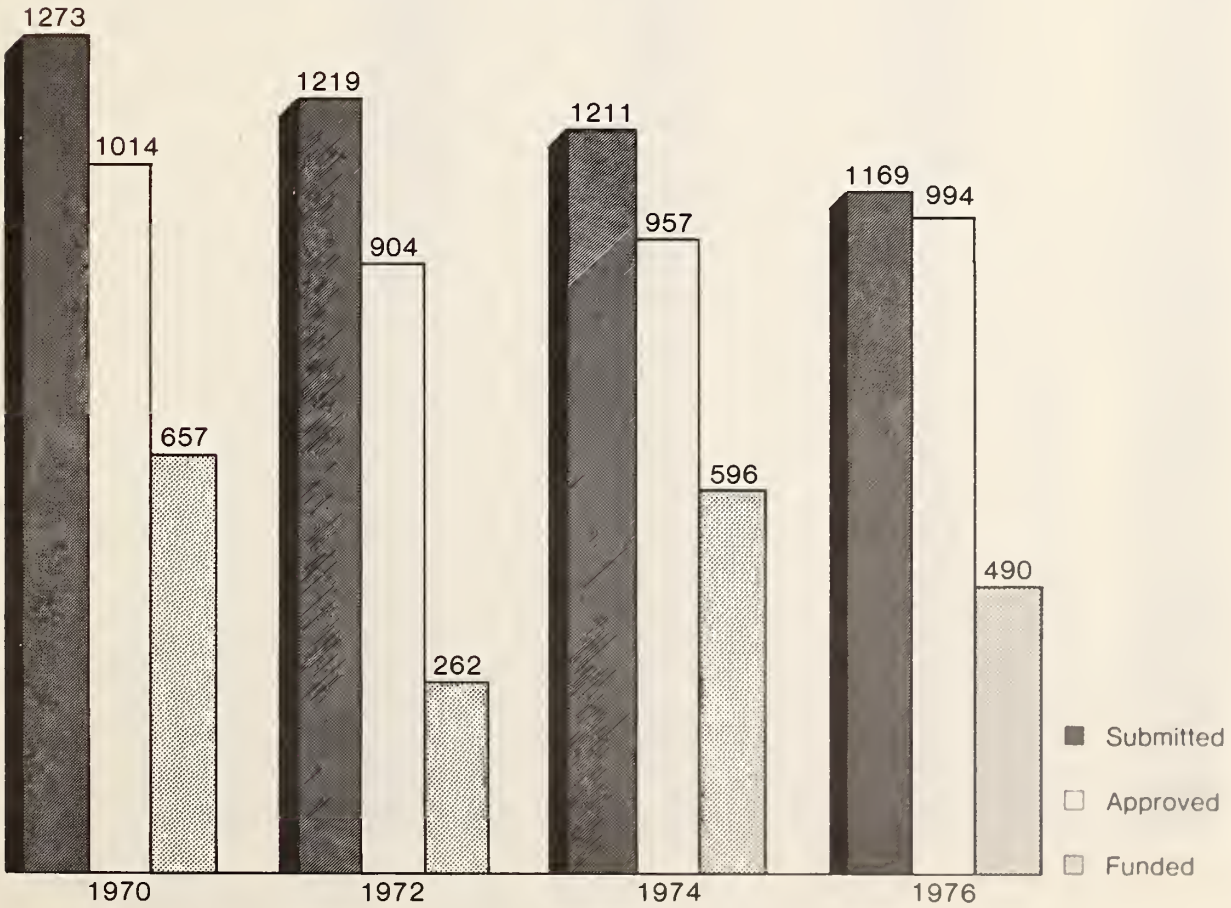
Over the past 16 years almost half the Nobel prizes in physiology and medicine have been awarded to neuroscientists. Floyd Bloom, President of the Society for Neuroscience, described the current situation in testimony before Congress:

> . . . the funds available for the support of this neuroscience research effort and for the training of neuroscience research personnel have deteriorated to disastrously low levels. At this time many of our members are unable to obtain support for their research programs and have essentially closed their laboratories.

The savings to society that would result from prevention and treatment of crippling neurological and genetic disorders fully justify increased research appropriations to the NINCDS and the other institutes and agencies supporting research on Huntington's disease and related neurological disorders. Targeted research on Huntington's disease must not be at the expense of productive research in other areas.

Huntington's disease has a scientific fascination quite apart from its medical and human importance. It first of all has a clear genetics, and so many diseases, certainly schizophrenia, do not have a clear genetics. It involves processes of cell death and aging, and the questions of what triggers and influences these processes are of utmost importance in science at the present time. In fact those processes also have ramifications for understanding cancer and aging. That is, why do cells die at a certain time and, conversely, why do they suddenly multiply beyond all bounds?

Competing Research Grant Applications Submitted, Approved and Funded by NIGMS FY 1970 — 1976*

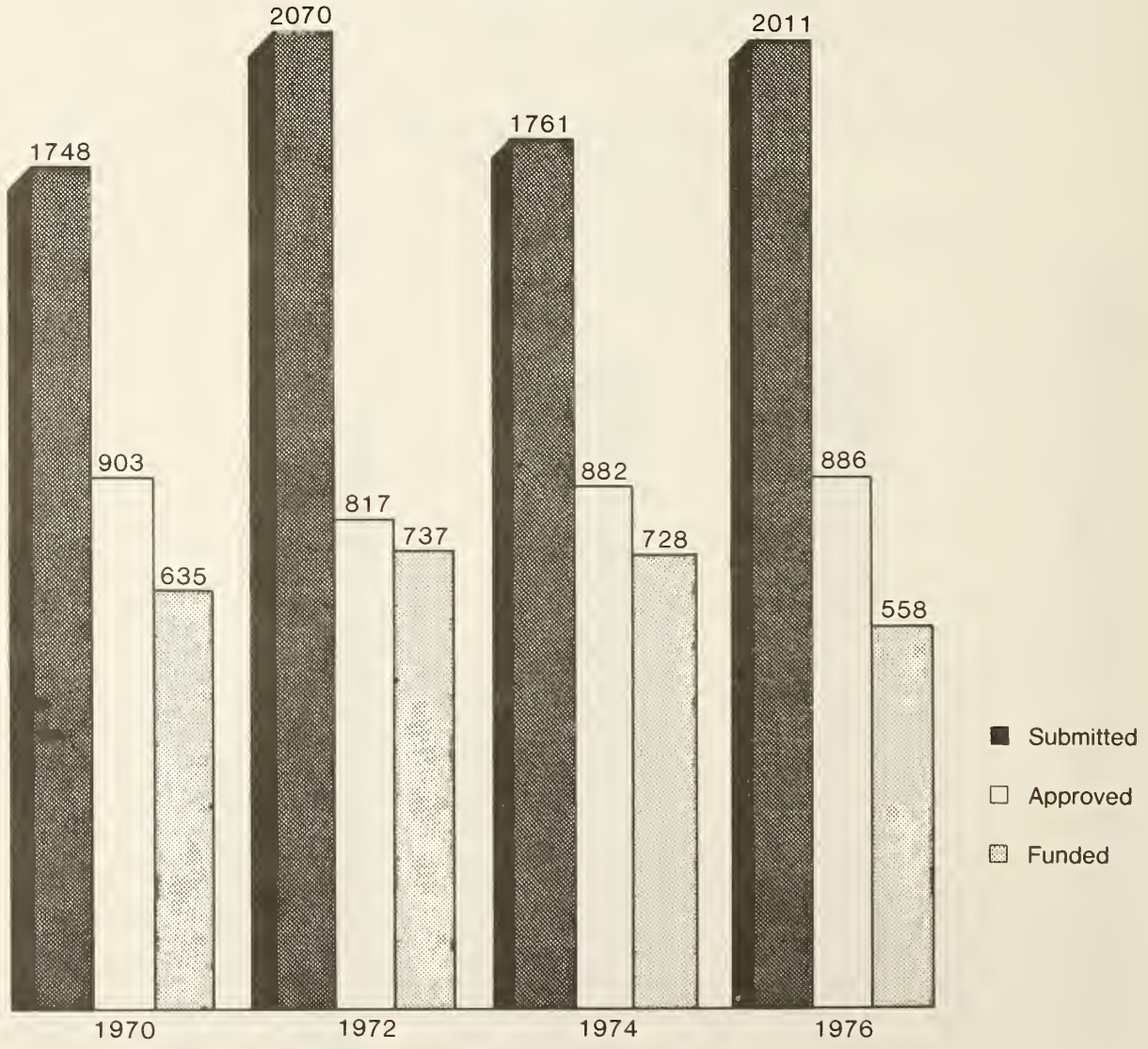


*Source: Statistics and Analysis Branch, Division of Research Grants, National Institutes of Health

The collaboration between biochemical, pharmacological groups on one hand, and electrophysiologists on the other hand, is not extensive enough. For example the relations between the cerebral cortex, the thalamus, the cerebellum and the striatopallidonigral complex have been relatively well studied by electrophysiologists. This is not the case in neurochemistry or pharmacology. This should be a privileged area of research in the future.

The Commission recommends that Congress increase appropriations to the NINCDS to $218 million for FY 1979, $245 million for FY 1980, and $273 million for FY 1981, to permit increased emphasis in research in the neurosciences. The overall amounts include the amounts specified for research in Huntington's disease in the Commission report. Furthermore, the Commission recommends that Congress increase appropriations to the NIGMS to $244 million for FY 1979, $259 million for FY 1980, and $274 million for FY 1981 to supply vital research in genetics.

Competing Research Grant Applications Submitted, Approved, and Funded by NIMH FY 1970 — 1976*



*Source: Grants and Contracts Management Branch, National Institute of Mental Health, Alcohol, Drug Abuse and Mental Health Administration.

My family has Huntington's disease. My great-grandmother was spoonfed in her bedroom; she had it. Her sister killed herself at the age of 24. My grandmother killed herself at 56. We assume she had it. Her brother "died" at 34 and another was bedridden for 15 years. My mother was severely mentally ill for many years; was in the State Hospital for seven years; attempted suicide; had shock treatments; was given the "air test" and diagnosed with H.D.; was in nursing homes three years; was cared for by me as a bed patient for three years; lost her husband; and went back to the nursing home two years ago. A burden to her husband and family for 25 years.

Her brother, Ernest, was mentally ill for 25 years and abandoned by his family of six. He is presently in a VA hospital. Her brother, William, had H.D. movements for 35 years and died in a VA hospital. Her brother, Harry, in ill health all his life, has been confined to a trailer home for five years (here on my farm). Her brother, Jeff, has lost a 24-year-old son with suicide and a 13-year-old girl died on the operating table, and one son is spastic and a state ward. My brother "died" at age 24 and two sisters have been in the State Hospital for awhile.

Including my brothers and sisters and all our children, we are presently 26 "at risk." If it averages 50%, as it has in the last three generations, that will be 13 more to suffer.

I needed help when I was ten years old and had to start keeping house and cooking and making the garden for my six brothers and sisters because Mom was always "sick." It seems to me my folk's life was entirely wasted through misinformation and no one to turn to. We are the first generation to know what's happening and be prepared for it. Our greatest hope is in research. We have donated brain tissue from Mom's two brothers. Our second greatest need is more localized medical assistance. Medical bills on all of this long-term care are astounding. I prefer the patient being kept home with assistance. The patients are aware of everything—remember, everything—and love their families, but lack judgment.

My name is Lynnett. I'm in the fifth grade. My father has had Huntington's disease all his life that I can remember. We had to stay by the house to watch him. We had to give medicine every night. Every year we go around for Huntington's disease. It can defect the brain. It can make you bad bad sick, think wrong, and it makes you mean. You pull off covers and pour water on people's head. It is a very bad disease. It makes nervous; you have to leave the house sometime. My mother would get griped out for being late. He lied on the couch day by day.

One day they might get a cure for it.

Ten years old.

It would take a book of encyclopedia size to write my personal stories of suffering, anxiety, poverty and desperation. But would anyone care? I am on welfare. I have signed over my home in order to have food on the table. We have no furniture and feel stripped of pride, dignity—not to mention my own health and state of mind.

I now have 6 children, ages 24, 23, 20, 19, 15, and 9. Where are they going to receive help? What encouragement can you give them besides looking forward to years of deterioration in a horrible mental institution which they don't belong and then sudden death.

Please! Please! do something now.

Better Drugs for All in Need

The Commission recommends that the President immediately appoint an interdepartmental Task Force to consider ways of encouraging and accelerating the development of new drugs to meet the urgent needs of small populations of patients afflicted by crippling or lethal diseases. The Task Force should include representatives of the Departments of Commerce, the Treasury, Health, Education, and Welfare, as well as members of Congress, representatives from the pharmaceutical industry, and consumers.

The pharmaceutical industry in the United States is a potent resource for the development of new drug treatments for diseases. It could be more constructively utilized with respect to small populations of patients who require new drug development to alleviate their suffering from crippling and lethal, but relatively uncommon disorders.

A pharmaceutical company encompasses a unique set of resources for the development and distribution of new drug treatments. A drug house has basic researchers who can create the actual compound needed, facilities for conducting toxicology studies, and mechanisms for performing clinical trials. A drug company also has the necessary distribution network to ensure that the new drug is properly marketed. Even if the NIH and university-based investigators were able to discover new compounds, perform the necessary toxicology and clinical studies, and clear the compounds through the Food and Drug Administration (FDA), they would still have to depend on the pharmaceutical houses to manufacture and distribute the drug. Sometimes drugs, proven effective in clinical trials, are stalled for considerable lengths of time at this critical juncture.

Marketplace Values

There are a number of complex factors which make drug development a costly and time-consuming process. These include steps to assure the safety and efficacy of drugs, reporting requirements, and a limited lifetime of patents during which companies can recoup their investments. The drug houses usually need assurances of eventual return on investments through a potentially large market before they will attempt new drug development. Considerations of the marketplace effectively bar small populations of patients from getting treatments they need.

The Commission held hearings with representatives of the drug industry, the FDA, the NIH, and prominent clinical investigators to clarify problems and explore innovative ways to mobilize the resources of private industry to spur new drug development. It was particularly disheartening to learn that almost none of the major pharmaceutical companies are involved in research on Huntington's disease. This is despite the fact that a drug which would be effective in treating Huntington's disease might also prove useful in the treatment of schizophrenia and other mental illness, dementia, and other movement disorders. *Even more ominous was the information that many pharmaceutical firms have curtailed or discontinued research on new treatments for any central nervous system disorder.*

Not Only Huntington's . . .

At the outset, the Commission sought to explore mechanisms of new drug development for Huntington's disease. It was soon apparent that the same factors which inhibit research for Huntington's disease hamper development of new drugs for all other small populations of patients. The Commission found this problem to be one of the most disturbing that it encountered in the course of its study. Neurological diseases and particularly hereditary disorders tend to affect only relatively small numbers, but in the aggregate they account for a major portion of the morbidity and mortality of this nation. Sickle cell disease affects between 25,000 and 50,000 patients in this country. Huntington's disease affects about 22,000 active patients with 44,000 more estimated at risk, a fairly large number for a genetic disorder. Taken singly, however, each of these killers and cripplers does not offer sufficient economic incentive to encourage

major new drug development efforts by the pharmaceutical industry. Must thousands languish and die for want of a vigorous pursuit of treatment?

Witnesses before the Commission proposed a number of remedies to the problem of new drug development:

- Change patent laws to extend the life of a patent for an unprofitable drug and thus assure the manufacturer a longer period to recover its investment.
- Provide tax advantages for drug development.
- Provide government subsidies to private industry or nonprofit organizations.
- Develop drugs through the NIH.
- Create a government-owned pharmaceutical company whose major concern would be drugs for limited populations (based on the Kabi drug company model in Sweden).
- Develop drugs in companies owned by foreign governments and liberalize regulations related to accepting data on compounds developed in other countries.

The most urgent need is to actively develop new drug treatments for crippling and lethal diseases. A related problem, once a compound has been developed, is to speed it to those in need. It has been estimated that the development of a single new drug will require at least 7 to 10 years. A possible solution to this problem is to draft special FDA rules for licensing limited-use drugs and maintaining such drugs on permanent Investigational New Drug (IND) status.

No branch of the Government currently has as its mission to actively initiate new drug development and shepherd treatment to the needy consumer, regardless of the profit involved. Consumers with uncommon diseases, who fail to arouse commercial interest in drug companies, have no representation in the private or public sector.

The Commission concluded that the many complex and intricate factors involved in arriving at the most equitable and effective solution to the problem of drug development called for special expertise and representation. The President therefore should appoint an interdepartmental Task Force consisting of representatives from the appropriate sectors of the Government, private industry, and consumers to begin work immediately to study the problem and make recommendations.

The Commission considers of top priority the development of new drug treatments for Huntington's disease patients and sufferers from other diseases affecting small populations who are all adversely affected by the current system of drug development. The Commission recognizes that careful study and implementation may require some time. *Therefore, the Commission urges that the Task Force be created immediately to remedy the situation in the very near future.*

One therapeutic approach to Huntington's disease is the use of drugs which mimic GABA to replace the presumed GABA deficiency. A focused effort at several research levels to develop effective GABA-mimicking drugs would have a fairly reasonable probability of resulting in agents with definite therapeutic benefit in Huntington's disease. The research technology, such as the ability to measure GABA receptors biochemically, is available. Since GABA is probably the major inhibitory neurotransmitter in the brain, drugs which mimic GABA might have application as sedatives, anti-anxiety agents, sleeping medications, and anticonvulsants. It is unlikely that such agents would be developed without some governmental input. Even though such drugs would have theoretical and practical utility which extends far beyond the boundaries of Huntington's disease, the attitude of drug industry might be that drugs for use in Huntington's disease lack commercial promise. A similar situation occurred in the early 1970s relating to a need to develop pure narcotic antagonists for the treatment of heroin addiction. Though this is not a commercially promising project, establishment of novel regulations facilitated a collaboration between industry, government and universities. This productive effort was organized by the Special Action Office on Drug Abuse Prevention in the White House.

The development of GABA-mimicking drugs is but one example of an approach to Huntington's disease which would have spinoff for many other diseases and in which specific governmental administrative maneuvers could facilitate progress. Such arrangements which might galvanize the drug industry into projects with both short and long term medical benefit to the public may be of relevance throughout the field of national health.

This patient has been treated on our Medical ward since 1971 at the per diem cost of $77.67 (as of April 1, 1977). Her father pays the hospital $250.00 per month; this represents roughly the cost of $137,000 of which the father has paid $24,000. No medication has been successful in giving the patient any comfort or relief of symptoms.

The emotional toll on the father is indescribable. The psychic pain of losing every member of his immediate family to a disease against which he is totally impotent and his only resource for care is as impotent as he is, is beyond expression.

Centers Without Walls

The Commission recommends that Congress appropriate funds and direct the Secretary of the DHEW to establish Centers Without Walls for research, training, and education in Huntington's disease and related disorders in accordance with the plan presented in this report.

The Commission is proposing a unique pilot program in research and management of Huntington's disease: Centers Without Walls. Each center would consist of investigators engaged in basic or clinical research at different universities throughout the country, and a staff and patients at a clinical research facility for Huntington's disease and related disorders. The clinical facility would be the hub of the Center Without Walls. Research investigators working on individual projects throughout the country would be separate, autonomous components. The investigators would be united with the clinical facility by their common research interests, by the frequent exchange of ideas through reports or meetings, and by common funding for the center. They would not necessarily be united geographically.

The Commission believes that Centers Without Walls can accomplish these major goals:

- Provide a clinical center of excellence where patients with Huntington's disease and related disorders will have access to an integrated program of medical, psychological, social, and rehabilitative services and can volunteer to participate in new drug trials and other experimental studies.
- Stimulate new research in Huntington's disease and related disorders by virtue of the heightened visibility a center creates and the availability of patient volunteers.
- Increase and quicken the flow of information among scientists with common interests but different research disciplines.
- Increase the rate by which new and effective methods of treatment and care developed in the laboratory reach the patient in need. The center would not only conduct research but also function as a demonstration and information center for rapid dissemination of the most recent and effective treatments, methods of care, and improved methods of diagnosis and prevention as they are developed.
- Provide a humanitarian focus in the community to serve as a source of hope and help to patients who have for too long despaired.

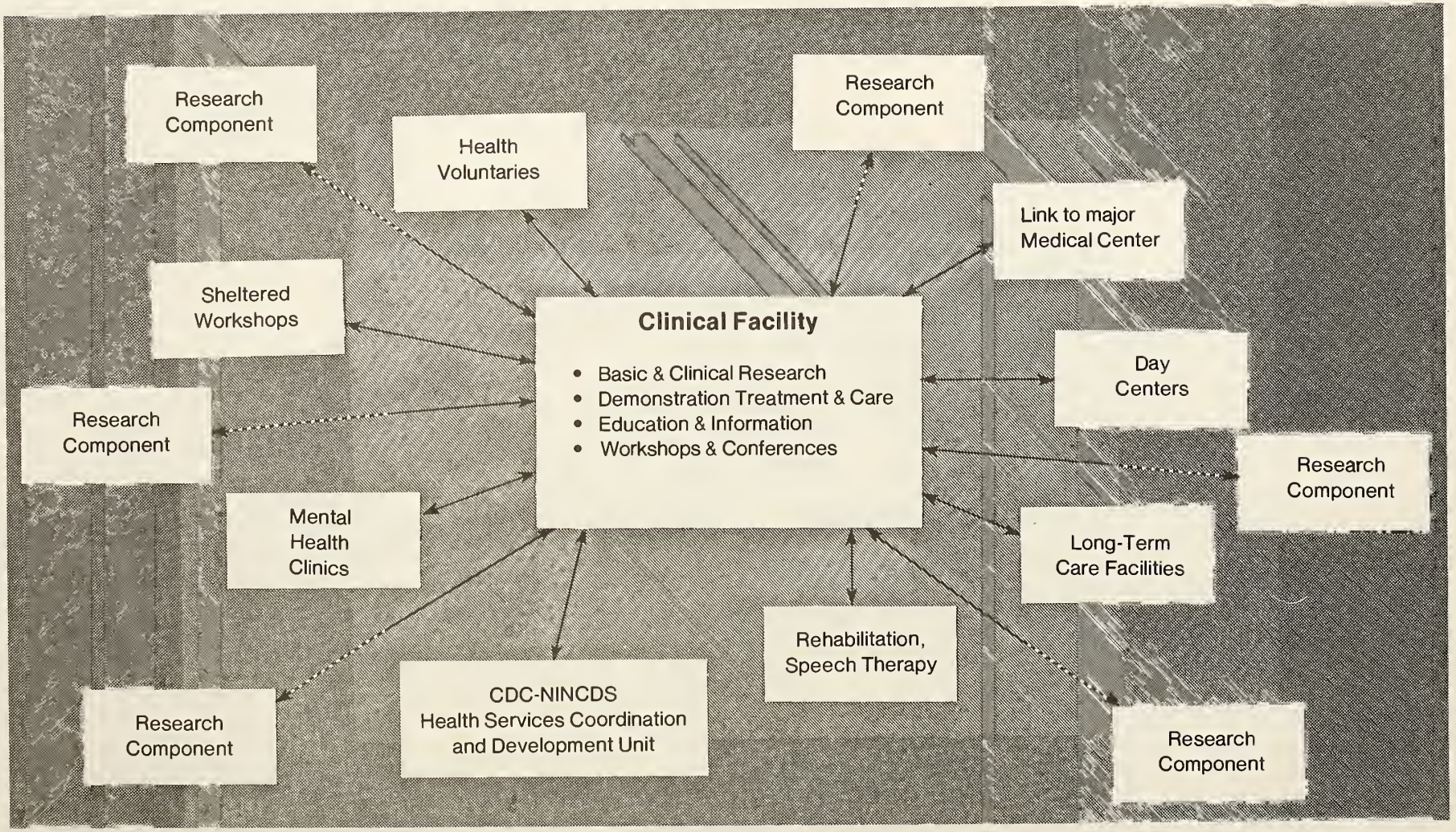
Not a "Traditional" Center

In developing the concept of Centers Without Walls the Commission has deliberately chosen to abandon the traditional idea of specialized disease centers. In the past this has led to the construction or utilization of buildings housing a staff of scientists, supporting personnel, equipment, and patients all under one roof. A major factor in the Commission's thinking was that *funds for Centers Without Walls would go primarily to support creative thinking and activity wherever they may be found, and not be spent on buildings and equipment. Center components do not have to be geographically contiguous.*

The result is a flexible plan. A Center Without Walls is designed to make optimal use of already existing resources and to develop new programs in cooperation with groups outside the center. Since each of the research components of the center is independent, it can be reviewed independently. Unproductive projects can be discontinued and promising programs can be incorporated.

Structure of the Center

As the hub of the Center Without Walls the clinical facility would serve as the administrative core of the center, with responsibility for coordinating Centers Without Walls projects. Its activities would include circulating information about ongoing activities and research findings, and generating educational materials for public and professional use. The administrative core would be expected to work with



A TYPICAL CENTER WITHOUT WALLS. A clinical facility would serve as a model demonstration, education, care, and treatment unit for patients with Huntington's disease and related disorders. The facility would serve as the administrative hub coordinating Center activities and working to develop needed services with local groups (including the proposed CDC-NINCDS Health Services Coordination and Development Unit). The facility might be a General Clinical Research Center in a medical center, a unit in a VA hospital, or a long-term care facility. The clinical facility would also be linked to basic and clinical investigators engaged in research in various parts of the country (*research components* in diagram).

community agencies in programs that could be developed in rehabilitation, counseling, and other forms of therapy. The administrative core would also develop community resources for intermediate and long-term care for patients. This would include long-term care facilities with a special relationship to the center to permit residents to participate in research if they so choose.

Research patients associated with the clinical facility would include Huntington's disease patients and patients with related disorders such as Parkinsonism, tardive dyskinesia, Tourette's syndrome or others according to investigators' interests. It is important that the clinical facility admit patients with several different but related disorders so that a variety of patients may serve as research controls for each other. It is also beneficial for patients to be in a mixed group where they can gain a perspective on their own condition, share experiences, and be mutually helpful.

The clinical facility need not be created *de novo*. It could make use of existing beds in a GCRC, a ward of a veterans hospital, or even a long-term care facility in the community. In all probability the facility would not need to duplicate the full resources of a major medical center as long as such resources were readily available for particular research programs.

Center Activities

A Center Without Walls would function as a laboratory for research in all phases of understanding, treating, and caring for patients with Huntington's disease and related disorders. The center, either at the clinical facility or through outlying projects, would support research exploring the underlying mechanisms that give rise to symptoms, and to research aimed at identifying gene carriers, or in search of methods of prevention and treatment. Critical experiments testing the efficacy of new therapeutics should be conducted. Participating investigators might be engaged in basic research on the brain or in genetics; they might be technicians developing new diagnostic equipment, or behavioral scientists developing psychological test batteries. Some of these projects may require access to patients or to tissues or fluids from patients. These investigators would draw upon the clinical facility to supply their need.

Research on the care of patients would also be conducted at the facility or at outlying projects. These studies would include research on the use of genetic or psychological counseling, on speech, rehabilitation and occupational therapy, and on programs of patient self-care, family involvement, and self-help groups.

In this way Centers Without Walls would become the vehicles for testing new findings before they are integrated into practice and for providing the most efficient and rapid translation of laboratory findings to physicians and health care personnel as well as to patients and families themselves. The center would have an outreach function to develop, study, and evaluate services in the community for patients and families. This work would be closely connected to the community "Health Services Coordination and Development Program." (See p. 51.)

Demonstration Care and Training

Some of the information on new therapies and care would be communicated directly through teaching and training programs conducted by the center. Training of both professional and paraprofessional personnel would be offered in techniques of research on neurodegenerative disorders and in the care and counseling of patients, at-risk individuals, and other family members.

I would like to underscore the comments that have already been made concerning regional care facilities. I think it's worth noting that where one finds active treatment centers, you are much more likely to find active research programs associated with it. When patients are scattered in care facilities, it's very hard to focus any kind of research program directed to a particular disease. From my point of view, as a researcher, a regional center would identify and localize a patient population for researchers interested in studying the disease. I think in turn — and I think it's important to say — that it would provide a way for those families who are interested in contributing to research to do so. Very often I think they feel frustrated by an inability to come in contact with people who are studying the disease.

Diagnostic Services and Data Collection

The center would provide complete and modern diagnostic evaluations of patients. It

would gather detailed clinical histories and obtain as full pedigrees as possible. This is especially important in cases presenting diagnostic problems.

Those records maintained by the clinical facility could form the basis of longitudinal studies of patients and persons at risk. Eventually, the records could be used for validating presymptomatic tests and for documenting clinical data and drug histories on patients who provide tissue for research studies.

Center Evaluation

In developing the concept of Centers Without Walls the Commission has deliberately sought to avoid the pitfalls that can beset the traditional concept of all-under-one-roof specialized disease centers. The Commission believes that all existing centers, as well as the proposed Centers Without Walls, should be reexamined and evaluated periodically to determine whether they should be continued, altered, or terminated. Many concerns are voiced about centers: they can become self-perpetuating beyond their useful lives; they are burdened with excessive staff and expensive underutilized equipment; or they become rigid and unable to accommodate to the currents of change in research. If a policy of periodic evaluation were routinely instituted, many of the problems could be resolved.

The Commission urges that two Centers Without Walls be established and that funding for the program initially be limited to a five-year period. During this time there should be a thorough evaluation of the components of the Center Without Walls to determine whether they should remain, be modified, or be eliminated.

Although additional authorization is not necessary for the establishment of the Center Without Walls, demonstration authority for the NINCDS would facilitate implementing the centers program and the dissemination of results of centers studies. (See Vol. II for authorization language.)

The development of centers for the treatment and research into Huntington's disease will, hopefully, produce resources and information for people who, like myself, are called upon for help. I need this. Those who turn to me need this even more.

When it was apparent that I had H.D., I began to lose everything. First came the separation, then a divorce. Along with the divorce I lost the house I had built, my car, my business, and above all, someone to lean on.

We were cut off almost entirely for several years socially. I was even told by one former friend, "I can't be seen with you. I will phone you and we will still keep in touch." This was almost more than one human could endure.

Her movements are unpredictable . . . The wheelchair is a problem for her, as with her movements she is easily bruised; they pad her chair, and she must be tied in so she will not fall out . . . She cries out, "I want to die!"

This disease seems bigger than the human capacity to love and have compassion.

I've been working in public psychiatric institutions for the past 30 years, both as a physician and as an Administrator. And during those 30 years the problems presented by degenerative diseases, including Huntington's chorea, have been continuing and frustrating. These patients are rarely welcome in nursing homes because of the difficult nursing and management problems they present. And unfortunately, our public psychiatric hospitals rarely have programs to adequately meet the needs of these patients. Our attempts to develop specific programs have not been successful because of lack of funding and because of the lack of trained staff.

Help is needed in patient care. My family learned mostly from experience about the best techniques in caring for my mother. Friends helped us in devising new ways of feeding and transporting her. This type of knowledge should be shared. Even trained personnel are sometimes not informed or experienced enough to handle H.D. patients. My mother was hospitalized during the week preceding her death. My father gave detailed instructions to the nurses about how and what to feed her. Yet when my sister returned later in the day, the nurses had not fed my mother. They said that she turned away when offered food. The nurses just hadn't understood, or perhaps hadn't cared, to take the time (often thirty minutes to an hour) to place each small piece of food deftly into the moving target of my mother's mouth. They didn't know that one hand, or even another person, had to steady her head while the other hand held the fork.

One of our greatest needs is a place where they can be taken care of properly—hospitals will not accept them and most of your better nursing homes will not accept them. If you are fortunate enough to get them in a convalescent hospital, it is just menial care and mostly the patients are restrained in bed 24 hours a day, so they lose what capabilities they do have.

During the last part of my husband's illness I often found him wet and/or soiled. No diapers were put on him. The air conditioner would be on and he would be cold. If they turned off the air conditioner, they would close his door and he would be too hot in the summer.

Mending Fractures in Health Care

The Commission recommends that Congress appropriate funds and establish the positions necessary to develop two pilot programs to improve linkages in community services and develop new services for Huntington's disease patients and families and those with related disorders. The Center for Disease Control (CDC) and the NINCDS should collaborate in initiating the project.

The fragmentation of services which could potentially benefit Huntington's disease patients and families, or others with chronic disease, is so extensive that the service system often frustrates the needs it was designed to satisfy. There is no *system of services* but, rather, a plethora of Federal, state, and local laws and programs. Dozens of different agencies and organizations administer these programs, each of which has different eligibility requirements and benefits, which also vary from state to state. A family shattered by the emotionally traumatic diagnosis of a fatal hereditary disease cannot cope with the maze of complex and baffling regulations with which they are confronted when they most need help. There is no *one place* to go for help.

This terrible illness has interrupted our lives like a tornado; we didn't know who to turn to. No doctor or hospital referred us to any organization who might have given us vital information as to how we handle ourselves in such a case as this. I'm at a loss as to what is available and how we can go about attaining aid and care when it is needed.

The Commission is not alone in finding that the fragmentation of health services is detrimental to good health care. The report of the Subcommittee on Health and Long-Term Care of the House of Representatives Select Committee on Aging begins with a discussion of the "proliferation and fragmentation" of services which they call a "national phenomenon." Many of the services required by the elderly are the same as those needed by persons with chronic neurologic disease. The reports of the Epilepsy and Diabetes Commissions also commented on the diffusion of services and the lack of centralized coordination.

Services exist in the community which could be adapted to meet the needs of patients with Huntington's disease and other chronic neurological disorders (such as vocational rehabilitation and Community Mental Health Centers). Other services simply do not exist but could be developed, such as group homes, respite care, and "Work and Recreation Day Centers."

The supervision of the victim becomes so constant that there is, literally, no time to spend on numerous phone calls or trips to remedy inevitable home crises or needs. One is not emotionally or physically up to tracking down resources.

In order to provide coordination and integration of services, the Commission is recommending two pilot programs of Health Services Coordination and Development to be carried out collaboratively by the CDC and the NINCDS. These programs would be organized around the needs of Huntington's disease patients and families. Other disease groups could be integrated as the program is expanded, and as they could be accommodated.

In addition to providing a rational linkage of existing services, the program would develop new services for individuals with similar needs. A public health professional would be assigned to a state or local health agency to work out ways in which a state or locally based community program could be adapted to the needs of Huntington's disease families and others with related disorders. The professional would be specially trained by the CDC and the NINCDS, and would be skilled in dealing with health professionals, voluntary agencies, and the general public. The coordinator's office would have a small staff (five persons) trained in relevant areas, including, if possible, new health care personnel: genetic associates, genetics social workers, genetics nurses and nurse practitioners. This staff would handle the daily management problems

of families while the public health advisor would be involved in developing new programs.

The initial duties of the coordinator and staff would be to assess the health resources relevant to the needs of patients with Huntington's disease and related disorders that are available in the area to be served. This could be a community or region. In the course of this inventory the coordinator would identify gaps in services and programs, and facilities which are redundant, underused, or in need of modification. This information would form the basis for a registry of resources used in making referrals and supplying information.

We were never given any plans or arrangements for appropriate care or services from the doctor, only to come back in six months, he could do nothing for her.

A single telephone number to the Health Services Coordination and Development office would serve as a "hot line" providing information to patients, professional health care providers, and the general public.

In addition to compiling information and serving as a central referral service, the Health Services Coordination and Development Program should include the following objectives:

- Serving as an advocate in the community, representing and making visible the needs and problems of this particular constituency. This is especially necessary when eligibility for rehabilitation or disability insurance programs is questioned, when access to convalescent facilities is denied, or when job discrimination occurs.

- Identifying, strengthening, and coordinating existing resources into a more rational and easily comprehensible service network with a systematic referral mechanism for treatment, services, and research. This entails developing and maintaining a resource registry as well as providing accurate eligibility information for appropriate programs.

- Developing additional community resources which may be beneficial to Huntington's disease patients and families and those with related disorders, such as day activity centers and mental health programs. This effort might involve integrating the services of a number of public agencies or voluntary groups with similar service needs. Where backing can be arranged, the health services coordinator can participate in the development of new programs such as respite or hospice care, which may benefit many disease groups.

- Providing coordination and facilitation for research projects, including the facilitation of tissue donation programs. The Health Services Coordination and Development programs should preferably be in areas where there are Centers Without Walls clinical facilities. There should be close collaboration between the two programs. The center staff can offer professional guidance and develop protocols to test the value of some services. The coordinators can carry out field tests of these services and give feedback to the center investigators.

- Developing and disseminating new educational materials for the patient, professional, and public, and providing training assistance to agencies and organizations giving service delivery at the community level.

The Commission believes that pilot programs in Health Services Coordination and Development can be of enormous value in providing services desperately needed by Huntington's disease patients and families and in developing and testing new programs.

These pilot programs should be established in two states for a three-year trial period, during which time they would be subject to ongoing evaluation.

The fact is that no place or setting exists in which such persons with chronic illness can find a positive and constructive environment which is sensitive to their illness.

I am the 70-year-old mother of three children, two of whom are in the advanced stages of H.D. These two are ages 43 and 44. I take care of them 24 hours a day, seven days a week, month in and month out. I feed them, bathe them, dress them, and do all the things they need done, even blow their nose and shave them . . . They drool continuously. This is offensive and hard to handle. My 44-year-old son wears a plastic diaper and a T-shirt only. I had to buy a chair that has a potty seat. He sits on it most of the day . . . I have five grandchildren by these two and they are looking the disease in the face . . . It is the worst kind of sickness I have ever seen.

I feel if there could be some hot line, or someone we could contact in our most crucial hours—which seem to be at night when all medication fails to help and the problem becomes uncontrollable, someone with a knowledge of H.D. could possibly tell us what to do, or at least talk to us, it would help, for we've had so many nights like this where neither of us got any sleep. Things are better during the day when she is more rational.

I cared for my son at home as long as possible, then placed him in a rest home. He left his room one evening to get something to help him sleep. The nurses put him back in his room and locked it with a coat hanger from the outside. He fell to the floor and after a time, managed to crawl to his bed and pull himself on it. One of the nurses told me that she and two of the other nurses watched him through an outside window. She said she didn't know how he ever managed it by himself. He told me about it the next day and asked me to come check on him about 11:00 o'clock as he had a fear of being locked in.

No one realized how devastating it was for me to have to take the step of going through commitment proceedings in the first place; and to have that followed by a series of case workers trying to throw her out!

Knowledge about resources is just not adequately disseminated. There's no catalogue in the library that I can go pick up that says, "H.D.," and which lists the people in the area who have the experience and the agencies which will work with the disease.

Chronic Disease Should Not Mean Impoverishment

The Commission recommends that the President, the Congress, and the Secretary of the DHEW ensure that the needs of the chronically ill are addressed in any program of National Health Insurance. Coverage of long-term care in such an insurance program should include the full range of medical, social, and mental health services required by the chronically ill.

The Commission recommends that Congress, the Secretary of the DHEW, through the Commissioner of the Social Security Administration (SSA), and state governments immediately take steps to correct deficiencies in the operation of the programs authorized under the Social Security Act, as amended, by adopting reforms specified by the Commission.

"Long-term care . . . is the most frequent cause of catastrophic expenses," according to the 1977 Congressional Budget Office Study, *Catastrophic Health Insurance*. For the family with Huntington's disease those costs can be impoverishing. First, the disease strikes in the prime of life. Beyond the costs of care, the family loses the income of the breadwinner or the homemaking services of the wife and mother—necessitating added homemaking expenses. Second, the course of illness is protracted—over 10 to 20 years. Third, the disease is hereditary. Not only must the family pay for the care of the adult patient, but children who are well now may also succumb—even while the parent is still a patient. Sometimes an elderly parent must assume the care of one or more grown children afflicted with the illness, at a time when he or she is faced with diminishing financial and physical resources. It is not at all uncommon for a single individual or family to be paying long-term care costs for several patients at once.

If you are not indigent when you walk in, you will be when you walk out — if you walk out.

Estimating the Costs

Variations in age of onset, severity of symptoms, duration of illness, and family circumstances make it extremely difficult to arrive at an average cost for health expenses in Huntington's disease. Witnesses before the Commission attested to the enormity of the financial drain, beginning with appointments with specialists to obtain a diagnosis (an arduous and costly procedure if an accurate diagnosis is not reached), to the costs of years of care in nursing homes or psychiatric hospitals.

Obstacles in Estimating Costs

A further impediment in accurately assessing the cost of Huntington's disease is that the total number of patients in the country is unknown. No nationwide or large-scale reporting of Huntington's disease is currently being done, and there are no recent surveys providing accurate data. The few statewide epidemiological studies of incidence or prevalence were undertaken in the 40s or 50s and have not been updated. There is also no organized data on where patients are being cared for, whether at home, in nursing homes, or state mental institutions.

Many Huntington's disease patients have been diagnosed incorrectly so that hospital records are often unreliable. Some patients undoubtedly have never been diagnosed at all. When the emotional or financial impact of the disease causes a family to break up, the patient may be lost to society and to statistics —living out life in a furnished room, wandering the streets, or abandoned to the back wards of a mental hospital as a demented or schizophrenic patient.

Representative Scenarios

To arrive at some estimates of overall costs, the Commission worked with the Huntington's

disease health voluntary organizations and the Health Services Research Center at the University of California at Los Angeles, to develop three representative case histories of Huntington's disease patients. In addition, the Commission studied data derived from a survey of 267 male and female Huntington's disease patients from across the country.

The UCLA figures were conservative estimates of *direct costs only*, primarily medical bills, drugs, or stays in private nursing homes, or state or Federal care facilities. The scenarios described three male patients, diagnosed at age 35 and surviving at least 15 years. Case 1 was a middle income male who was able to stay at home throughout the illness. Case 2 was a low income male who in the course of illness was divorced, was eligible for Medicaid, and spent five years in a state mental hospital. Case 3 was a man who could afford to stay in a variety of private intermediate care facilities for 10 years and then, finances depleted, was moved to a state mental hospital. The range of costs was approximately $64,400 ($3,220 a year) for Case 1; $118,000 ($5,900 a year) for Case 2; and $233,800 ($11,690 a year) for Case 3. Assuming a conservative estimate of 15,000 Huntington's disease patients in the United States (a prevalence rate of 7 per 100,000), those figures would yield an annual cost in the range of $110 million to $125 million.

Minimum Costs Only

The cost figures the Commission has estimated are conservative and minimal. They do not reflect the impact of the disease on the family. Often the psychological stress and physical exhaustion imposed by the disease lead to alcoholism, drug abuse, depression, or other emotional disturbances in patients and other family members. The costs of the disease invariably include excessive clothing and laundry bills, and replacement of broken furniture or other household items. The costs given do not cover counseling or psychotherapy for at-risk individuals or other family members. Nor do they include any rehabilitative measures which might improve the patient's condition, such as speech or occupational therapy. They do not include the costs of special foods, wheelchairs, hospital beds, or any of the devices handicapped persons require.

The Costs of Health Care Resources Used by Three Typical Huntington's Disease Patients*

	Physicians	Medication	Psycho-therapy	Home Health Assistance	Private Residential Care	State Institution	MISC. General Hospital, Genetic Counseling, Legal Services, Psychiatric Social Worker, Sterilization	TOTAL
Scenario 1	$5,925	$2,142	$9,500	$44,250	—	—	$2,600	$64,417
Scenario 2	$1,825	$2,888	$2,550	$5,200	—	$100,995	$4,750	$117,988
Scenario 3	$5,175	$900	$5,200	—	$119,000	$100,375	$3,150	$233,800

*Information from A. Fink, J. Kosekoff, and C. Lewis, "A Study of the Resources Available to Huntington's Disease Patients, Families and Health Care Providers," UCLA Health Services Research Center Report No. HS-2015, July, 1977.

Lost Income

Moreover, the costs omit lost income. Data on 267 Huntington's disease patients pooled from members of the Commission's work group on Behavioral Changes and Treatment provided rare clinical information never before gathered on a sample of that size. The average age of these patients was 47. The average age at the time of diagnosis was 42, with initial symptoms appearing four years earlier. All but a handful had been wage earners or homemakers at the time of diagnosis. At the time of the survey, however, 87 percent of the wage earners were unemployed. Assuming that a person of 47 would normally work at least 15 years more and earn the U.S. average annual income of approximately $10,000, a Huntington's disease patient or the family stands to lose at least $150,000 in income if the breadwinner becomes ill.* That loss is also society's.

The Middle Income Gap

The loss of income (or housekeeping services) when combined with the high costs of long-term care imposes a financial burden which is particularly hard on middle income families. To qualify for financial aid or health benefits under existing programs of the Social Security Act, the family may have to be reduced to the poverty level (which may have the effect of discouraging the healthy spouse from working). If income criteria for eligibility are not a barrier, criteria requiring that the patient be a wage earner for a specified period of time can be an additional impediment. Moreover, the health benefits under these programs are woefully inadequate in supplying the needs of the chronically ill patient.

Available Care Is Expensive Care

The plight of middle income Huntington's disease families is shared by other middle income families who must pay the catastrophic costs of long-term care. The most common form of long-term care is institutional care in a nursing home or other residential facility. Yet institutional care is the most expensive form of long-term care. *The average cost of a year's stay in a nursing home is projected to be $12,700 in 1978.*

Yet 35 percent of the families of disabled individuals in America had incomes of less than $3,000 a year in 1975; 70 percent had incomes of less than $7,000 that year. In the case of Huntington's disease, a family must often pay costs of nursing home stays of 15 years or more. At the 1978 rate this amounts to nearly $200,000.

Despite the enormous discrepancy between income and expenditures, it is the families of disabled individuals who bear the brunt of the financial burden. Family out-of-pocket funds accounted for more than half the payments for long-term care in 1975.

Projections show that government expenditures for institutional long-term care will decline in the next few years, while private insurance payments will remain minuscule. Who pays for the rest? The families of the disabled.

The public testimony of Huntington's disease families revealed how they manage. Individuals took out bank loans or second mortgages. They sold the family car, pawned the furniture, or moved to smaller quarters in a less desirable section of town. Those who could work got jobs, even if it meant quitting school or leaving the patient at home alone or with younger or more elderly relatives.

It is a matter of no small pride for some families that they have been able to manage this way. For others, the overwhelming costs prove too much. The household is reduced to the subsistence level, and only then, ironically, does the family become eligible under existing government programs.

I have been advised by our County Department of Social Services to get a divorce, thereby making my husband a dependent of the State and he would be eligible for Medicaid.

There is another alternative to financial ruin—divorce. In that way the well spouse can be spared bankruptcy and save for the care of the patient or for children. In turn, the patient, now indigent, becomes eligible for care and services as a ward of the state.

**If multiplied by an estimated 6,750 wage earners who develop Huntington's disease (assuming 10 percent of the estimated 15,000 total cases are juveniles, and half are women who may not be employed) the total loss of income to the country over 15 years would be $1 billion.*

All these alternatives are unacceptable. Exhaustive, superhuman efforts . . . reduction to a poverty level . . . divorce—fly in the face of the ideals of a stable family life, a decent standard of living, and humane care for the ill.

What is needed is a program of National Health Insurance which will provide comprehensive coverage for the chronically ill. While such a major program is being developed, reforms of existing financial aid or health benefit programs under the Social Security Act are mandatory to correct present inadequacies.

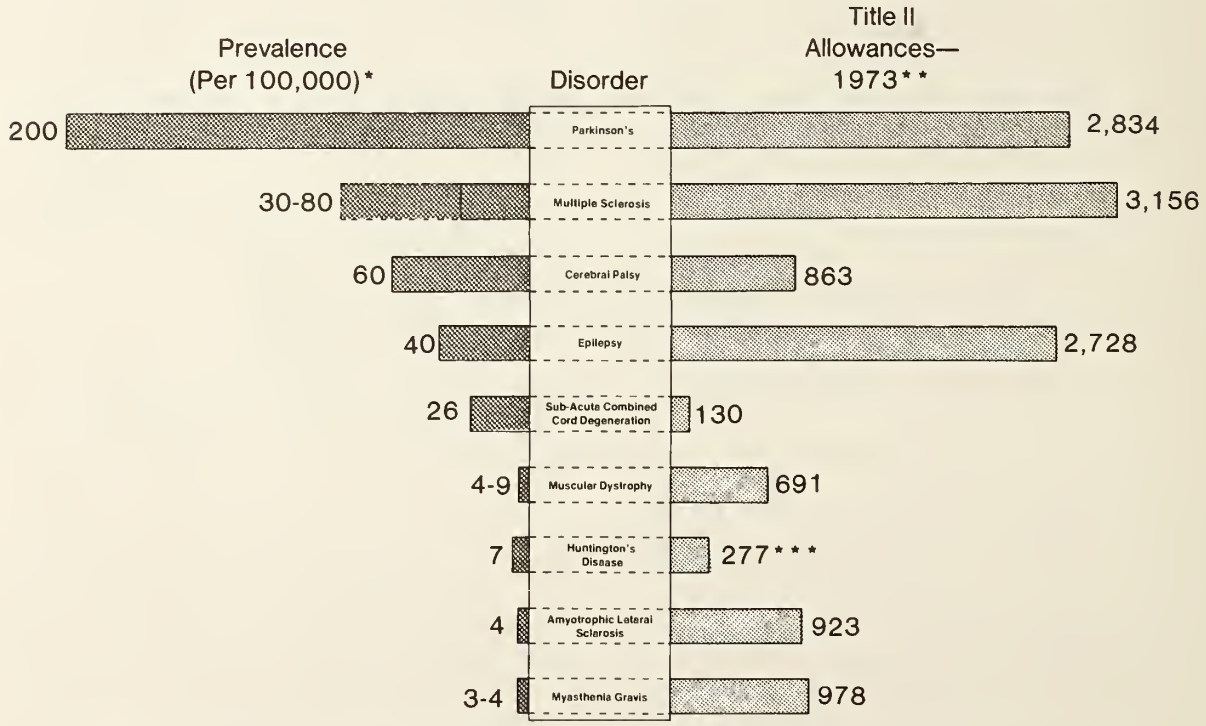
Financial Aid Programs

Disability Insurance — Title II

The Disability Insurance Program, Title II of the Social Security Act, provides financial assistance to disabled workers. To be eligible, an individual must satisfy criteria concerning the total time worked and the recency of the work history. *Women with chronic disease who have been homemakers are not covered because their labor in the home has not been for wages.* Huntington's disease patients may have difficulty meeting the requirement for recency of work. Psychological changes, which are often a part of the insidious onset of the disease, may appear many years before the diagnostic abnormal movements. These psychological changes may seriously impair job performance and regularity. There may have been long periods of unemployment or frequent job changes. There are no medical guidelines specifying how these early behavioral changes are to be considered in the determination of disability. *But these changes should be recognized as part of the disease process by those determining eligibility.*

Huntington's Patients Underserved—Evidence suggests that many Huntington's disease patients who are eligible are not receiving Disability Insurance. The most

Number of Disability Claim Allowances Under Title II of the Social Security Act (FY 1973)



*L. Kurland, J. Kurtzke, and I. Goldberg, *Epidemiology of Neurologic and Sense Organ Disorders* (Cambridge: Harvard University Press, 1973).

**Elmer W. Smith, Associate Commissioner for Program Policy and Planning, SSA, to Marjorie Guthrie, Chairwoman, Commission for the Control of Huntington's Disease and Its Consequences, 28 June 1977.

***Data include Friedreich's ataxia and spino-cerebellar degeneration in addition to Huntington's chorea.

recent data supplied by the SSA shows that the number of allowances for disability insurance for Huntington's disease patients in 1973 was very low in comparison with other neurological disorders, based on estimated prevalence rates.

These data indicate that those who develop Huntington's disease either do not know of this benefit or, for some reason, are being disqualified.

Supplemental Security Income — Title XVI

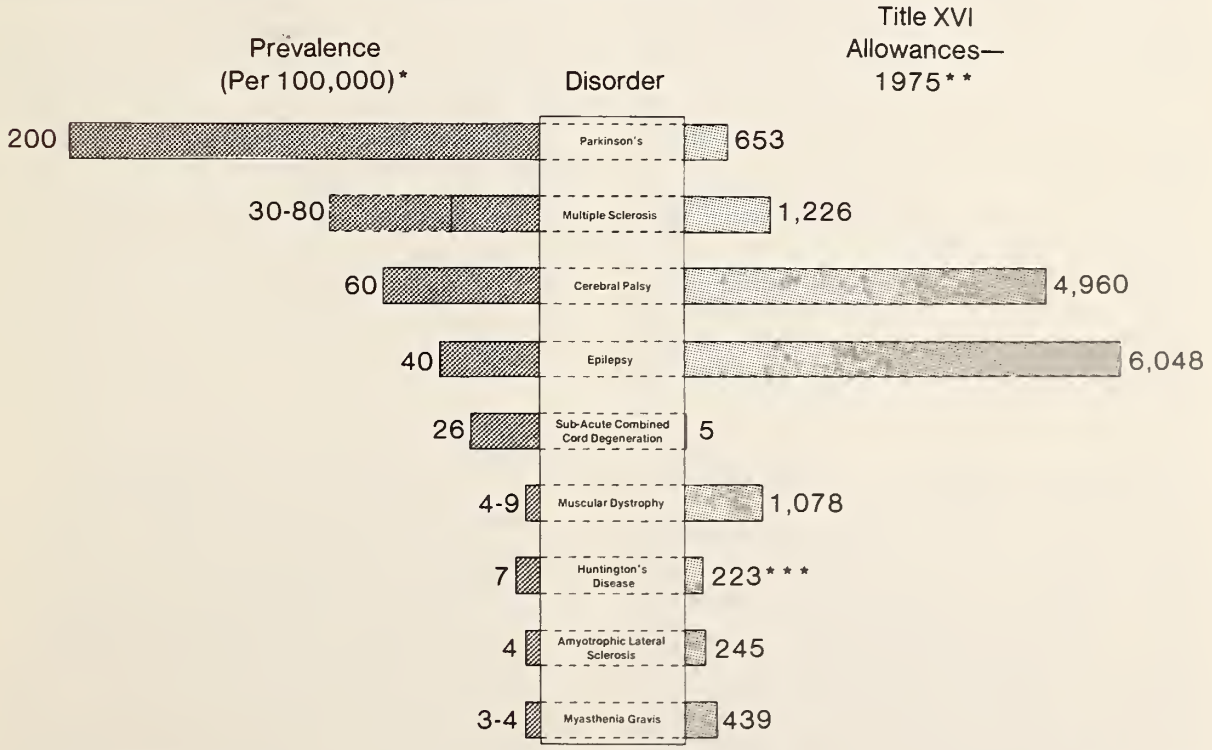
Federal, state, and local cash grants are available to the needy aged, the blind, and the disabled under the Supplemental Security Income Program (SSI), Title XVI of the Social Security Act. To be eligible an individual must have an income near or at the poverty level and possess minimal assets.

A principal advantage of SSI is that it entitles individuals to Medicaid, social services (Title XX), and other benefits. For Huntington's disease patients these benefits are even more crucial than the income supplement. The dilemma the family faces is that if income goes above the low limit set by the state, not only is SSI lost, but also the Medicaid and other benefits. Medicaid will pay some of the costs of long-term care. If the family loses Medicaid and then must assume long-term costs, it will soon be reduced to poverty again.

In any case, a family receiving SSI has no way to save for the health or educational needs of other family members, including at-risk children. The threat of the loss of Medicaid and social service benefits deters family members from working to earn a decent living. Worse yet, the regulations effectively encourage divorce so that the patient can continue to receive care while the healthy spouse and other family members try to gain financial independence.

A fairer determination of the total family income is needed. Moderate amounts of funds placed in trust for children's education, for future medical needs, or for some other

Number of Disability Claim Allowances Under Title XVI of the Social Security Act (FY 1975)



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purpose related to reasonable anticipated family needs should not preclude SSI eligibility.

To counteract the work disincentive effects of SSI eligibility requirements the Commission urges *that an individual who has been determined to have a disability that will last longer than 12 months and will result in death, and has met the family income criteria for qualifying for Medicaid and social services as established by each state, be entitled to continued Medicaid and social service benefits even if total family income should later become equal to 115 percent of the state's median income for a family of four.*

The Need for Strong Leadership

Eligibility for disability insurance and SSI (when based on the criterion of disability) is determined by the same state agencies using the same medical criteria. Again, the figures indicating the number of Huntington's disease patients receiving SSI seem disproportionately low in comparison with other disorders.

It is possible that outmoded criteria and methods of determining disability account for the low number of allowances for Huntington's disease patients. An independent report of the Comptroller of the United States in August, 1976 found that "state agencies have not received strong and active leadership from the Social Security Administration (SSA) in determining who is qualified to receive disability payments." The study pointed to weakness in the medical criteria and ineffective quality assurance activities of the SSA.

More vigorous leadership and guidance from the SSA might help secure more equitable treatment for victims of Huntington's disease and similar disorders.

Health Care Financing Programs

In addition to income assistance programs, a number of public and private plans cover health care expenses. Unfortunately none of these programs is adequate to the needs of the middle income family faced with the castastrophic expenses of chronic care. Some programs apply only to low income families. Others do not begin to supply the range of services needed by the chronically ill. Still others exclude patients with Huntington's disease on the grounds that it is an untreatable illness or requires only custodial care.

Medicare

Individuals receiving disability insurance payments for a continuous two-year period are eligible for Medicare. But the Medicare program is an adjunct of the acute hospital system and does not meet the needs of the chronically ill. Services are intermittent or part time, and are only provided during or following an "episode" of acute illness.

Home Health—Medicare provides some home health benefits, but these concentrate on skilled services for the acutely ill, not on health-related or basic services for the chronically disabled.

Skilled Nursing Home Care—Medicare will cover the costs of staying in a skilled nursing home, but the length of time is limited to a maximum of 100 days per benefit period after three days of hospitalization—clearly of limited value to chronically ill patients.

Medicaid

Medicaid is the principal source of Government financing of long-term care. It is the only Government program which provides protection against chronic catastrophic costs, but the program is only available to the indigent or to those who are reduced to near poverty in the course of financing long-term care. A study by the Congressional Budget Office, *Long-Term Care for the Elderly and Disabled*, describes this unfortunate process:

> Only by "spending down" to income levels that make them eligible for Medicaid payments do they get government assistance for long-term care. *This impoverishment of the disabled is suggested by the fact that 69 percent of nursing home residents have incomes under $3,000 and that over 47 percent of nursing home patients whose costs are paid by Medicaid were not initially poor by state definitions but depleted their resources and became qualified as "medically needy."* (Emphasis added)

The Medicaid program in 31 states extends coverage to the "medically needy"—individuals whose income is too high to be eligible for Medicaid, but not high enough to meet medical costs. In general the medically needy are still relatively low income individuals. Coverage is

also highly erratic. In a study of one state program in 1975, less than 5 percent of those eligible received benefits. This is an example of just one of the many ways the Medicaid benefit package varies from state to state.

The designation "medically needy" is of great help to many people and should be adopted by all state Medicaid programs and vigorously implemented.

Institutional Care—Although coverage under Medicaid theoretically extends to a range of health services, the program is, in practice, oriented to skilled services in an institutional setting. Over 98 percent of all Medicaid long-term care expenditures in 1975 were for nursing home care.

Home Health Care—Home health services, including domestic chore and personal care services, have been covered by Medicaid since 1970. The extent to which these services are available is questionable, however. One state alone accounted for 70 percent of all Medicaid home health payments in 1975. Expenditures for home health services were only $70 million in 1975, less than 2 percent of all Medicaid long-term care expenditures.

Social Services — Title XX

Title XX of the Social Security Act provides funds to the states to develop social services for low income individuals and recipients of cash assistance programs. Homemaker and chore services, day care, and foster care are available for those eligible. However, only $66 million was spent on such services for the disabled in 1975.

The fourth position lasted less than two weeks because the company's personnel department had initially overlooked LuAnne's honest admission of having HD. She was subsequently ruled unacceptable for group insurance coverage and thus unemployable by company personnel rules.

Private Insurance

Private health insurance plans rarely pay for long-term care costs. Nor do they usually cover long-term home health care and social service expense. Moreover, the individual who is already ill or who admits a family history of hereditary chronic illness is unable to obtain insurance, finds the policy cancelled, or is unable to renew a policy if it expires. In some cases, companies explicitly deny coverage for Huntington's disease; other policies stipulate that coverage does not apply to "non-treatable" illness or to custodial care.

"Out-of-Pocket" Funds

Following a nationwide trend, private out-of-pocket funds comprise the bulk of payments for long-term care of Huntington's disease patients. Eighty-eight percent of private source payments are out-of-pocket. There is no price tag on the cost of care given by friends and relatives, although these expenditures—in money, time, and energy—are enormous.

The company we sought [insurance] from, we answered truthfully upon the advice of the doctor. There was no known reason why we should not be insurable. But the notation in his records of a risk of Huntington's disease—and they refused any kind of insurance. What a blow! The next worry was that we may have been put on a black list and not be able to receive it anywhere.

National Health Insurance

Huntington's disease patients and families share with other Americans the frustration and despair of not being able to find the level of care suited to their needs, nor of being able to afford such care if it exists. (See p. 65, "Nothing to Do and No Place to Go: Care Which Meets the Need.") In common with other disabled persons, Huntington's disease patients and families have discovered that regardless of their best efforts to remain financially independent they cannot be self-sufficient. Long-term care costs grind them down to poverty and the welfare level.

The Commission finds it imperative that a program of National Health Insurance be developed which addresses the problems of the chronically ill. It concurs with Dr. Julius Richmond, Assistant Secretary for Health of the DHEW, in insisting that the National Health Insurance plan must go beyond issues of financing to address realignment of priori-

ties and development of "an integrated, coordinated system of care with quality control and cost effectiveness." *National Health Insurance must cover the full range of medical, social, and mental health services.* Special programs in the Department of Housing and Urban Development (DHUD), the Department of Transportation (DOT), and other agencies outside the DHEW can provide supportive services for the chronically ill and should be included in a program of comprehensive long-term care.

The Commission appreciates that National Health Insurance requires major study and careful development and that a special panel, the National Health Insurance Advisory Council, is studying the issues. *Accordingly, the Commission urges that the Secretary of the DHEW ask the National Health Insurance Advisory Council to provide a special report on the inclusion of a chronic illness program in the administration's health insurance proposal, and assign a special work group to develop a plan for the chronically ill.*

The Commission recommends that the Secretary of the DHEW through the Commissioner of the SSA immediately take the following steps to correct deficiencies in the operation of programs authorized under the Social Security Act, as amended:

New medical guidelines for determing disability under Title II (Disability Insurance) and Title XVI (SSI) should be issued, incorporating the most recent findings about the nature and course of Huntington's disease. These guidelines should be distributed to SSA regional and district offices and to state vocational rehabilitation offices.

A program of centralized review of claims for allowances under Title II and Title XVI on the basis of disability due to Huntington's disease should be initiated to assure quality control and uniformity of decisions.

Regulations regarding Title XVI should be modified to exclude, in determining resources of individuals with permanent disability leading to death, moderate savings placed in trust for specific stipulated purposes, such as education or future medical needs of children, whether placed in trust before or after application for benefits under Title XVI.

Income eligibility requirements should be revised to permit continued Title XIX (Medicaid) and Title XX (Social Services) benefits to individuals with permanent disability leading to death, until family income exceeds 115 percent of the median family income for the state.

The Commission recommends that Congress consider new legislation or amendments to the Social Security Act to address the needs of women who become disabled. Coverage under Title II (Disability Insurance) of the Social Security Act should be extended to persons who have been homemakers and whose labor in the home has not been for wages.

The Commission recommends that all state agencies administering Social Security programs under Title XIX (Medicaid) of the Social Security Act adopt the designation "medically needy," and ensure, through vigorous implementation, that all who are eligible receive benefits.

There is something wrong with a society that says that a person with a debilitating disease for which there is no known cure, must not only carry the burden of his fate, but must also be financially penalized for having that illness. Although the H.D. person does receive Medicare, that only covers the doctor's bill and medication. Nursing home care is denied. The only way a family can survive is to go on welfare. Why should a man or woman be forced not *to work to support his family because a spouse has an irreversible illness requiring constant care beyond the means of the workingman? Instead of the ill person being put on welfare, an entire family is kept from being active, productive citizens.*

As it stands now, one must be very poor or wealthy to get quality health care.

I would either have to take him home or I would have to place him in a state institution, or if I wanted a private facility, I would have to dissolve—liquidate—every bit of finances that we had. Every bit of income up to, I believe, $2,500. And I would be allowed $300—under $300—a month to live on. Therefore, I would have to quit my job and could no longer earn my salary because it would be over and above the required amount. I could not afford to place my husband into the private facility or even the state facility or even the city, because I couldn't afford $1,200 a month. I still had, at the time, a daughter at home, that was still going to school and I had all the other personal needs of any human being who is alive and well and trying to live as best they can. So the financial situation had become extremely impossible.

As I cared for my husband for almost thirty years, I was unable to build up my Social Security. Now at age 78, I am going home to care for my daughter.

Another thing, I have to get special car insurance, even though I have never had a ticket or an accident, because my husband has Huntington's disease. They say there is always the chance he may try to drive. He has a difficult time just walking—he'd never try to drive a car again.

Nothing To Do and No Place To Go: Care Which Meets the Need

The Commission recommends that a continuum of care services and facilities be available to Huntington's disease patients and others with chronic or disabling conditions, to permit individuals to function at an optimal level commensurate with the degree of impairment. Local Health Systems Agencies and State Health Planning and Development agencies should ensure, through their Health Systems and Annual Implementation Plans, that health provider organizations develop such services and facilities.

The Commission recommends that Congress appropriate funds and designate the NINCDS as the lead agency in the implementation of pilot programs for patients with Huntington's disease or other neurological impairments in: research and demonstration of long-term, institutional care; respite care, arranging for scheduled short-term stays in community health care facilities; and Work and Recreation Day Centers providing social, recreational and rehabilitation services.

Patients with Huntington's disease and other chronic illness can lead lives of dignity and productivity, they can be more self-reliant and learn self-caring skills, given services, aids, and training to complement their own resources. Many of the services patients need are everyday aids to living: meal preparation, assistance in grooming, transportation, companionship. Other needs include social services and the professional skills of counselors or therapists.

Those services are very different from the medical or surgical interventions associated with acute illness. Yet they are vital in maintaining the health and well-being of chronically ill patients. Cures for many diseases must await future research discoveries, but practical means of care and treatment are available which can make the difference between lives wasted in costly institutions and lives spent at home and in the community enjoying a measure of independence and activity.

Unfortunately, the needs of patients with long-term illness have been consistently overlooked in the provision of health services. Inadequate planning, financing, and manpower development have resulted in systematic neglect of long-term care. The neglect continues in spite of a 1970 DHEW Task Force report that "the failure to address the problem directly distorts the operations and inflates the cost of the medical care programs.

Comprehensive Long-Term Care

What is needed is a program that permits flexibility in the range of services, facilities, and personnel to supply the physical and emotional needs of chronically ill patients over the long course of disease. In stabilized conditions such as some cases of cerebral palsy or epilepsy, the level of care required may stay the same over the course of the patient's lifetime. In progressive degenerative conditions such as Huntington's disease, the patient's needs inevitably change and grow over the course of illness, moving toward the need for skilled nursing care and medical supervision in a more protected environment.

The only place the government agencies would allow her to go was to a MENTAL HOSPITAL! And we watched her with hollow eyes as she spent six months in the geriatrics ward of a mental institution, 105 miles away from her family and her home. Can you imagine what that did to all of us? Can you imagine what it did to a lady who has all her wits about her (better than I do) and is being punished because she can't walk by herself or feed herself or speak clearly? My father took her home again and tried to struggle on by himself. At least he and my mother have regained some measure of dignity.

The variety of needs requires a *continuum of services* suited to the needs of the chronically ill at any degree of impairment or stage of disease. The level of care should range from minimal once-a-week housecleaning services or a daily telephone call, to the fully protected environment and skilled care needed by a patient in the end stages of illness.

Home Care

People generally prefer to stay at home in familiar surroundings and with the family when they are chronically ill. But home health services are virtually unavailable. The SSA estimates that up to half a million disabled persons between 18 and 64 could benefit from home care, but no more than 60,000 or 70,000 individuals received such services in 1975. Medicaid, which provides over 77 percent of government support of long-term care, spends less than 2 percent ($70 million) on home health care.

A broad program of home care would enable patients to stay in their preferred environment for as long as possible. A home care program is also likely to be more healthful and less costly in the long run. It is estimated that between 20 and 40 percent of the nursing home population could be cared for at less intensive—and less expensive—levels if alternatives were available in the community. At present, home health and day care, and personal care homes or sheltered living arrangements, can only meet about 25 percent of the demand for services. In contrast, the demand for institutional care, which accounts for 90 percent of all long-term care expenditures, is only 20 to 30 percent.

With home care assistance, these people often could be reasonably supported in the homes they have struggled throughout their healthy years to maintain.

Without home care assistance, the family becomes exhausted, both mentally and physically. Then hospitalization is resorted to. This really costs. Instead of paying these huge medical bills, you could drastically reduce this by providing an attendant, part-time helper, or money to hire home help. Everyone would benefit: the patient, the family, the insurance carriers, the hospitals, and the community at large.

In the case of Huntington's disease, patients might be able to remain at home 10 years or more after diagnosis if adequate help were available. The problem lies in finding that care. Throughout the public testimony families stressed the need for help in caring for relatives at home. If the family breadwinner has Huntington's disease, usually the healthy spouse must work. If the homemaker is affected, additional help is needed in managing the home. Most families cannot afford domestic help, private nurses, or companions—especially when finances are depleted by medical and other expenses. As a result, families are sometimes forced to leave the patient alone—a situation as depressing and demoralizing for the patient as it is worrisome for the family. All too often the family resolves this dilemma by institutionalizing the patient, even though institutional care is inappropriate.

Moreover, the fragmentation of long-term care programs, of which home health care is a part, only adds to the bewilderment of patients and families. *As many as five different Federal agencies run six separate programs that fall under the heading of long-term care.* It takes more effort to devise a home care schedule than to place the person in an institution:

> To provide home care, it might be necessary to obtain income assistance from the welfare department, homemaker services program, and health care from the Visiting Nurses Association, with each agency insisting on its own review of the patient's eligibility. If one element of the "package" should fall through, the patient could not remain at home.—*Long-Term Care for the Elderly and Disabled*

Home health services and other basic services defined under Medicaid and under Title XX of the Social Security Act should be available to qualified disabled individuals regardless of income levels. Fees can be set on a sliding scale so that middle income families could avail themselves of these services at moderate cost.

Congregate Housing

Many chronically ill individuals live alone without a family to care for them. Yet their condition does not warrant institutional care. Not infrequently, Huntington's disease patients are alone because the stress of the illness often leads to the disruption of home and family. Where does the solitary patient live? The Commission learned during public

testimony that Huntington's disease patients live in furnished rooms or apartments where they burn themselves when they cook, and spill or drop things on the floor. Some live on the streets. Some commit petty crimes so that they can stay in jails. But they maintain a pride, a need for independence. Nursing homes, veterans hospitals, state mental institutions are not for them—yet.

Some form of group housing would answer the needs of those patients. Such facilities are called congregate housing, sheltered living arrangements, domiciliary care facilities, personal care or group homes, according to the specific plan and range of services provided. Typically, a personal care home is a residence in the community accommodating a group of individuals under the supervision of a resident attendant. Meals, housekeeping, and social activities are provided, as well as transportation and access to community services. Usually it is beneficial to group individuals with a variety of stable or progressive conditions. The main purpose of the home is to be a refuge for the Huntington's disease patient and others who have no home or family.

Estimates vary widely on the actual number of such units available, but they appear to meet no more than 20 to 30 percent of the estimated need.

Several programs of the DHUD provide financing for congregate housing. The programs supply construction money or rent subsidies. But "lack of interest on the part of private developers and absence of guarantees by state and local service agencies to provide meals and other services" have meant that only 22,560 units were funded under these authorities.

Work and Recreation Day Centers

To relieve the burden of boredom or loneliness, chronically ill patients need someone who can provide minimal supervision, services, and a degree of companionship. Such care is generally considered custodial, however, and is not reimbursable under health plans. Home health agencies provide specialized or skilled services for shorter periods of time and do not have personnel who can spend much time with a patient. As one young woman with Huntington's disease who stays at home all day with her father, also a Huntington's patient, said: "You think all day about the end—how it will come."

Day care for young children has become a well-established tradition in America, enabling many mothers to work. Centers for the elderly are becoming more popular. Between infants and the aged, however, there is a large population of young adult and middle-aged neurologically impaired individuals who have nothing to do and no place to go. These people desperately need to be involved with society outside their family.

My husband is certainly not ready to be put into an institution. We do not want to put him in an institution. It is most important at this time, when he can still function, to keep him physically and mentally alert and active . . . We want him to be with us and we want to be with him. But how can you see this happen to a person and see them lay in bed from morning until night . . . and keep telling you that they're going crazy for the lack of something to do? And there's no place to take him and no facilities for him to go to. If there were only places where he could go for the day. And come home and return to us at night and be with us. When they're put in institutions, nothing is done for them.

A Work and Recreation Day Center would provide a pleasant environment for individuals outside the home. They would be with others and have opportunities to learn, enjoy new skills, even work productively in a sheltered workshop. Such a center could be established in association with non-profit organizations or health voluntary agencies to provide mutual assistance. Staff members could include personnel trained under the Comprehensive Employment and Training Act.

The social and recreational aspects of the center should emphasize the importance of maintaining physical fitness, good diet, and proper hygiene. There should also be access to therapies, such as speech therapy, and to psychological counseling. As with group homes, a Work and Recreation Day Center should serve individuals with a broad range of illnesses and handicapping conditions.

Even more of a problem for me than finances, however, is BOREDOM. Since the chorea movement has become quite pronounced, no one will hire me and my only recreation is going to the library and CCHD meetings.

Under the social services program, an estimated 36,000 disabled adults received "day care"—rehabilitation and social services at a center during the day. *With the need for such services estimated at between 1 and 1.5 million persons in 1976, it is clear that major expansion of the program is imperative.*

Respite Care

An unbroken schedule of caretaking exhausts even the most loving and patient family and can lead to the institutionalization of a patient before it is necessary. This could be forestalled if families were assured that they would have time off or time away from the patient at regular intervals.

"Respite" care is the solution. It is just what the name implies—a way of providing respite from the constant burden of caring for a chronic patient and the constant dependency of the patient on the family.

Respite times can be provided by homemaker or domestic chore personnel working in the home on a regular part-time basis. Respite times can also be provided by paid companions. These individuals need not be skilled nurses, but they should know first aid techniques, be able to prevent choking, for example, and they should assist the patient in personal hygiene. Their main function would be social and supportive.

Of course, life in our household is far from easy. I find that a teenager, a pre-teen and an H.D. parent just don't mix. We live a life of constant compromise, and of trying to do anything for a quiet life! There is not a great deal of laughter in our house. Sometimes we spend a whole day of peace. This is something to be thankful for because the next three days may be filled with anger. Really we could all use a vacation, but how can we? This is an area in which some kind of Government-financed respite care would be of great help.

A program of respite outings or trips for patients has been effectively instituted in Australia. A program of regularly scheduled outings for day, overnight, or longer periods could be arranged as part of the activities of a Work and Recreation Day Center, or be included in programs arranged in cooperation with health voluntary organizations.

Another alternative for respite care involves hospitalizing patients for short periods on a regular basis. This has been tried with great success in England. In one community, chronic neurological and geriatric patients were hospitalized every two weeks for 48 hours. (Provision was made for longer stays if necessary.) During their stays, patients were given medical check-ups.

Respite hospital programs could easily be established in the United States given the large number of vacant beds that "deinstitutionalization" or overconstruction have made available. A wing of a community hospital could be converted for a respite care program for geriatric or neurologic patients. Such a program would have major advantages:

- Patients would be checked periodically by medical professionals to permit early detection of complications.
- Patients might be able to remain in the family home much longer—past the time when they might normally have been institutionalized.
- Families would benefit from the relief from stress and strain.
- Underutilized beds in hospitals in the community would not go vacant.
- A variety of architectural designs could be tested to see which would be most suited to the special needs of patients with movement disorders, and senility and dementia.

While there are costs involved in hospitalization, the amounts involved in weekend stays as opposed to the costs of full-time institutionalization would make it cost-effective.

Respite care is not widely discussed as one of the elements of long-term care coverage. *Yet its inclusion in any comprehensive plan of long-term care is important, particularly to address the problem of the burdens faced by family members in caring for patients at home. Provision for respite care and its financing should be included in comprehensive long-term care programs.*

Mental Institutions, Nursing Homes, and Intermediate Care Facilities

Given the difficulties of maintaining a patient at home and the lack of availability of

group homes, Work and Recreation Day Centers, and respite programs, it is not surprising that many patients with Huntington's disease are institutionalized. On the basis of public testimony, 83 percent of Huntington's disease patients were then living or had at some time lived in nursing homes, VA hospitals, or mental institutions. In an ideal continuum of care there is a place for institutions, but these must be facilities which provide humane and appropriate care. Most existing nursing homes, psychiatric hospitals, or state institutions are inadequate or inappropriate for Huntington's disease patients for the following reasons:

- **Age and Sex.** Huntington's disease patients are middle aged men and women. The median age of nursing home residents is 82, and 73 percent of the residents are women.
- **Safety.** Huntington's disease patients are subject to falls. Most institutions have no architectural protections against this hazard. Patients with movement problems are often forced to abandon independent locomotion and are restricted to wheelchairs or tied in bed.

She rolled between the mattress and the side rails on the bed and suffocated to death.

- **Staff Training.** Most staffs of nursing homes and mental institutions are not equipped to understand either the physical or the emotional problems of the Huntington's disease patient. Nursing homes in particular often cannot handle the patient's psychological difficulties. If the patient is placed in a mental hospital, however, the presence of schizophrenic or other seriously disturbed individuals is terrifying to patient and family and adds an additional stigma to the illness.
- **Patient Reaction.** The bizarre movements of Huntington's disease patients often frighten other residents who ostracize and isolate the patient. Patients are

A Continuum of Care in the Community: Settings and Services

Settings

family home | group home | sheltered workshop | work and recreation center

day hospital | intermediate care facility | skilled nursing facility

respite care facility | state hospital | mental hospital | hospice

Services

medical care | genetic counseling | emotional counseling/crisis help

vocational rehabilitation | financial and legal services | housing

transportation | homemaker and chore assistance | meals | personal grooming

companion/social services | recreational and occupational therapy

speech and physical therapy | respite care | nursing care

aware that others find them frightening and upsetting.

Many nursing homes refuse to admit a patient with the diagnosis of Huntington's disease—if they have heard of the illness. If they do admit such patients, many are also quick to transfer the patient out.

These conditions would be enough to discourage placing a Huntington's disease patient in a general nursing home or mental hospital. In addition, there are very real dangers of abuse or substandard conditions. The subcommittee on long-term care of the Senate Special Committee on Aging estimated that at least 50 percent of nursing homes are substandard, with violations including "negligence leading to death and injury, unsanitary conditions, poor food or poor preparation, hazards to life and limb, lack of dental care, eye care, podiatry, misappropriation or theft, inadequate control of drugs, reprisal against those who complain, assault on human dignity, and profiteering and 'cheating the system.' "

To correct these abuses will require a thorough revision of Federal regulations for nursing home facilities to create uniform, easily comprehensible and enforceable standards of care, with guarantees to assure that all nursing homes meet these standards.

Comprehensive Health Care Planning

To develop a positive program providing a continuum of care requires awareness of the need and systematic planning. The record to date has been inadequate. Health officials at all levels of government and provider organizations have not addressed the needs of long-term care sufficiently in either policy decisions or planning documents. This is in spite of the fact that between $12 billion and $13 billion were spent on long-term care in the United States in 1975—11 percent of all health expenditures.

A minimal start toward developing an adequate program of comprehensive long-term care, as cited in *The Forward Plan for Health*, would be "the development of national guidelines regarding medically appropriate home health services for various patient conditions as a screening manual, in concert with PSRO guidelines." But recognition that a full range of services and facilities is needed must precede the development of guidelines. As a first step, an inventory of the needs must be made to encourage "the development of various levels of care on a geographically integrated basis." (*The Forward Plan for Health.*)

The National Health Planning and Resources Development Act of 1974 is purported to be "the foundation for coordinating the diverse health care systems and for the systematic development of health resources at the state and local levels," (*The Forward Plan for Health*) in preparation for National Health Insurance. Plans formulated under this Act must encompass the full extent of existing and needed community resources; they must provide a variety of living facilities and arrangements, enriched social and rehabilitative services, new modalities and settings for long-term care, and full utilization of existing facilities and resources.

The Commission feels that it is vital to improve the quality and levels of long-term health care. Each Health Systems Agency must make provision for services and facilities that are consistent with the health care needs of patients in respect to delivery, amount, and timing of services, in accordance with the National Health Priorities of Section 1502 of the National Planning and Resources Development Act of 1974.

The Commission recommends that Congress appropriate funds and designate the NINCDS as lead agency in the implementation of the following pilot projects, all of which should be coordinated with Centers Without Walls when possible:

1. *A research and demonstration project for the long-term institutional care of Huntington's disease patients and others who are neurologically impaired.* Innovative programs in long-term care using staff specially trained in neurological nursing should be provided. New architectural and interior designs should be explored to determine how best to provide a comfortable and hazard-free environment which is also conducive to keeping neurologically impaired patients mentally alert and oriented.

2. *A respite care program for Huntington's disease patients and other patients living at home.* This program would arrange for regularly scheduled short-term stays for patients

in an underutilized community hospital or other health care facility. Patients would receive medical care and have access to other services during their stays, and families would be free of caretaking responsibilities at these times.

3. *Work and Recreation Day Centers for Huntington's disease patients and other chronically ill or disabled persons.* The centers would provide social and recreational activities, rehabilitation programs, a sheltered workshop, and other social or therapeutic services. Staff could include personnel trained under the Comprehensive Employment and Training Act.

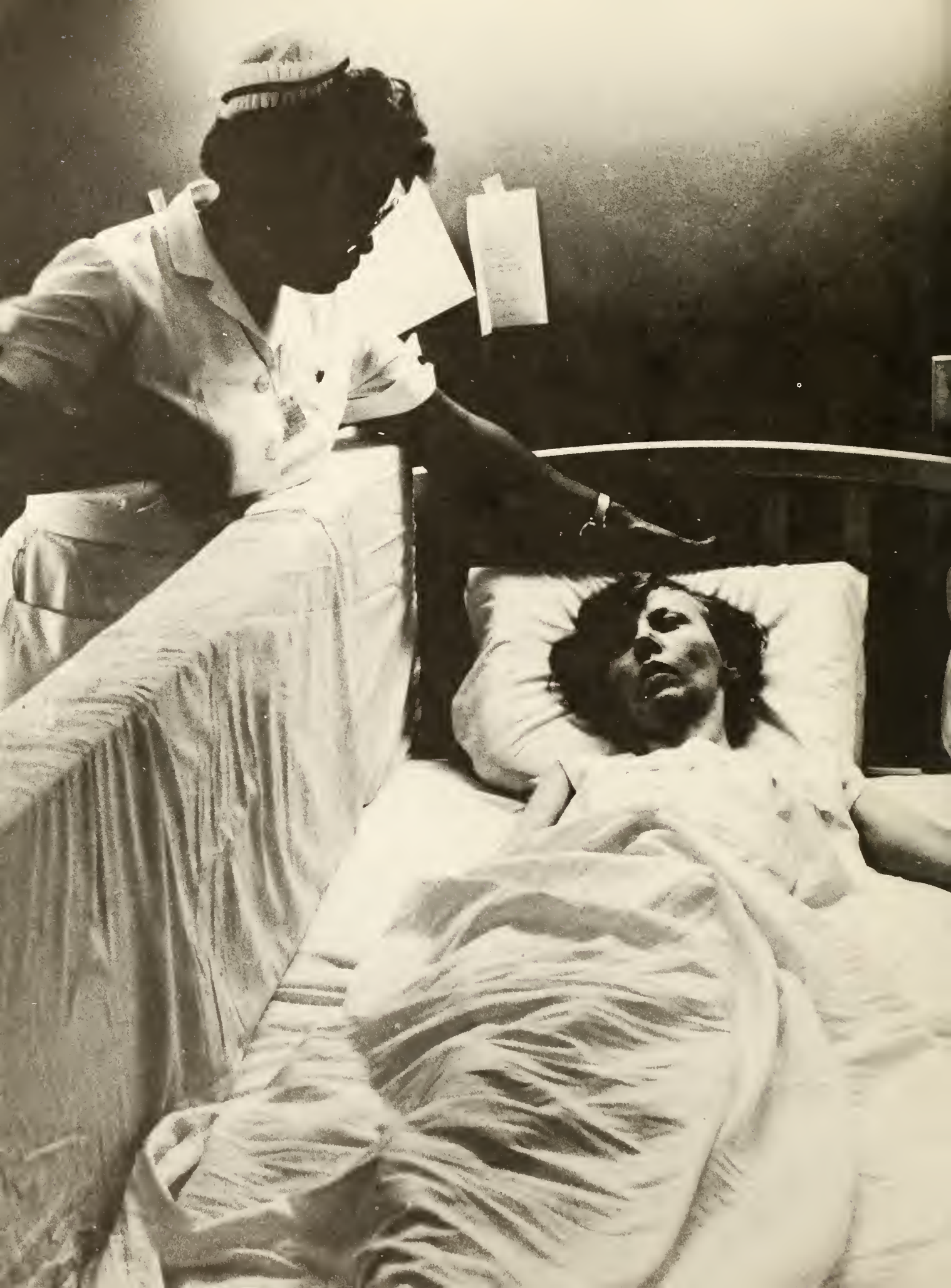
One of the most important things that we need is some kind of a day care center where victims with H.D. could go during the day, with crafts and other activities and therapy.

I didn't want to put Mom in a nursing home, but that was all that was left for me to do, and everytime I have been in there, they have had her tied down in a chair. I did not tie her at home; she is not an animal.

But they said they are short-handed and could not watch her all the time.

There are plenty of summer things for the severely handicapped or retarded and plenty for normal children, but nothing for the in-between child.

The nursing home only gives him a sponge bath (when they feel like it). Sometimes he's still in bed at 2:00 o'clock in the afternoon. Most of the time his face isn't even wiped, food still smeared all over. He can't talk anymore, can't call or ring the light, when he needs help. They don't put a catheter on him because they say they can't get one to fit him. They didn't even know how to get him from the wheelchair to the bed when he came in. He fell out of bed about two weeks after he was there because they didn't even put the sides up. They only seem to pay attention to people who can be up and about and can talk. The helpless ones get nothing.



Creating New Jobs in Health Care

The Commission recommends that jobs and services in long-term health care be developed under the auspices of the Comprehensive Employment and Training Act (CETA) utilizing public assistance jobs. New health care personnel pilot programs should be established in several states.

The disabled and those with chronic debilitating disease need a rare kind of health care provider—someone with time to give. The American medical care system is one of the most sophisticated in the world. It is geared for fast action and dramatic cures. But there are some diseases and disabilities for which time, attention, diversion, and even affection are the only treatments. Huntington's disease is one.

People with multiple impairments of mind and body need multiple sources of help. This help can be provided by individuals with training in many fields or by interdisciplinary teams of professionals and paraprofessionals working together to provide comprehensive care.

Some chronically ill or disabled persons need basic services more than skilled assistance. Ironically, unskilled help is often more difficult to obtain. Fewer agencies provide it; almost no insurance plan reimburses for its cost.

Huntington's disease patients and families need the traditional skills of trained health care specialists. Equally pressing, however, is their need for people who can relieve the psychological, social, and practical burdens of the illness.

The Forward Plan for Health

In recent years attention has focused on the need to ensure quality of care and reduce unnecessary institutionalization. This has led to a new emphasis on developing a variety of alternative care settings and services. *The Forward Plan for Health,* for FY 1978-82, prepared by the Office of the Assistant Secretary for Health for the Public Health Service, stresses the importance of providing appropriate care through other modalities:

> . . . inadequate attention has been paid to the development of non-institutional forms of long-term care. The concept of care outside an institution is broader than has generally been acknowledged. It is not limited to home health services alone, but includes a wide array of in-home support and maintenance services . . . as well as such new modalities or settings for long-term care as day care centers and day hospitals . . . Clearly, an effort will have to be made to increase access to noninstitutional care. In addition, however, a strategy will have to be developed to assure the quality of noninstitutional care.

It was evident in Commission testimony that many families affected by Huntington's disease try to maintain the integrity of the family for as long as possible before institutionalizing a patient. This is particularly true when patients are young and are parents of young children. These families need a variety of services to minimize stress and promote family life. Likewise, patients who live alone but who are not ready for institutionalization could have many productive years if community services were provided to help them.

"Better Jobs and Income" Program

President Carter's new Better Jobs and Income Program could help provide those services. The program has expanded certain provisions of the CETA and calls for the creation of 1.4 million public sector jobs, some of which are in the health services. This program is well suited to explore the development of new health care personnel while providing meaningful employment to large numbers of the unemployed in this country. The CETA program could be used in pilot tests of comprehensive long-term health care and social support programs. Training, supervision, and evaluation programs can also be developed and tested. Pilot tests using individuals trained under a CETA program in a number of areas can yield much needed information on the demand for services, on patterns of service use, and on patient and provider satisfaction.

The variety of jobs and services which could be developed under the program includes the following:

Domestic Chore and Homemaker Services

- Household chore and maintenance work
- Preparation of meals; laundry service
- General housekeeping

Transportation Services

- General "dial-a-ride" services to provide the equivalent of door-to-door taxi service for homebound or institutionalized patients
- Taking patients to and from medical appointments, sheltered workshops, etc.
- "Recreation rides"—taking one or more persons on outings
- Taking research patients from home care facilities to participate in research projects (to give blood, etc.)
- Transporting biological samples and research supplies from laboratories to appropriate depots for shipping

Companion Services

- Companions—visiting in homes or institutions on a regular, part-time, or full-day basis
- Day care for young children at home with ill parent while spouse works
- Reading to visually impaired persons
- Accompanying patients to religious services, movies and stores
- Telephone assurance—someone who will always answer on a "hot line"
- Shopping services—for homebound or institutionalized individuals—groceries, clothes, prescriptions, etc.

Therapist "Extenders"

These individuals would be trained by professional therapists and follow established guidelines in their work. Activities would include making home calls to ensure that therapeutic regimens are being followed. Family members are often discouraged in efforts to keep a patient active, taking medication, carrying out other programs to promote health, and keeping involved in family matters.

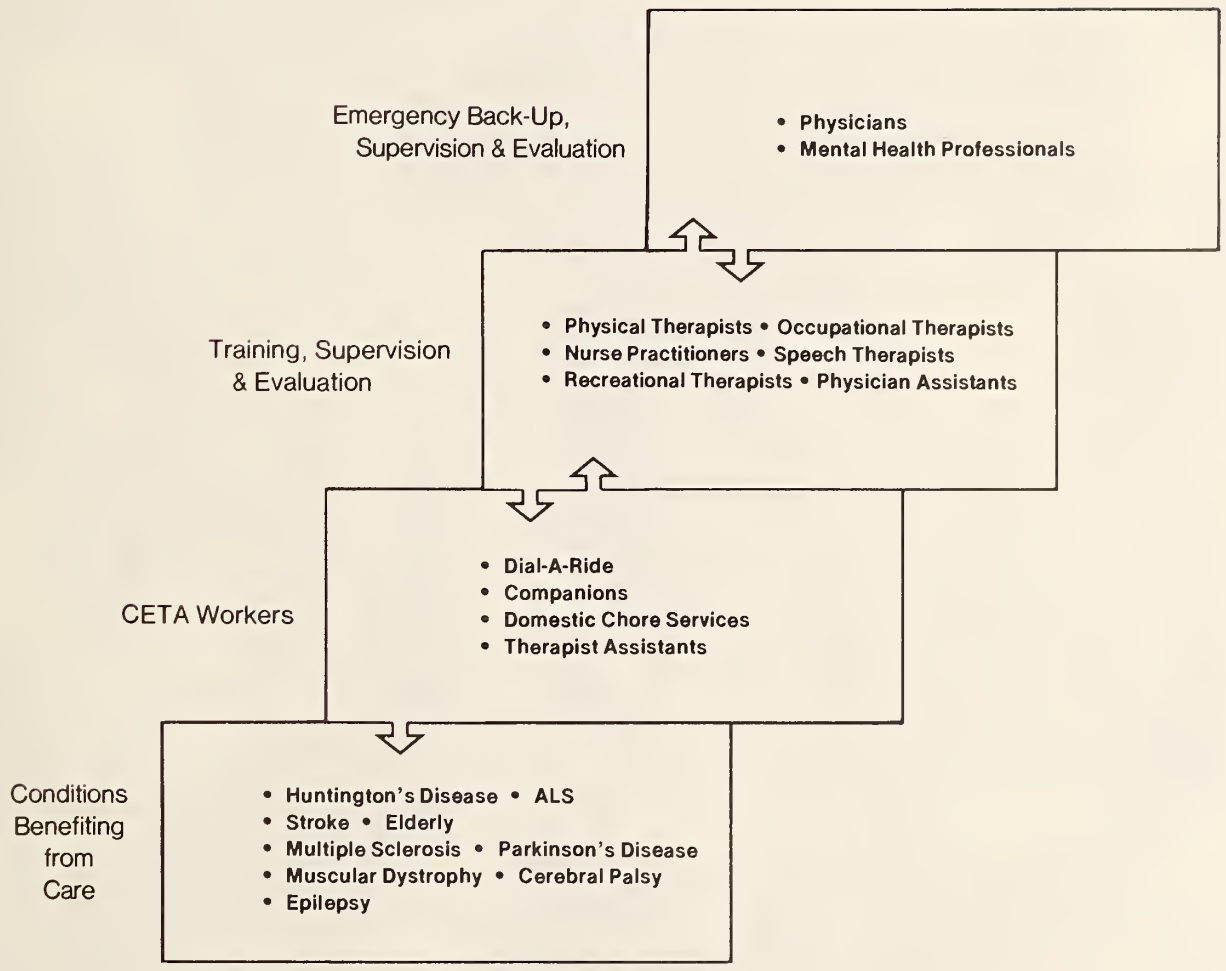
- *Physical therapist assistants* can make home visits to lead exercises.
- *Occupational therapist assistants* can teach crafts and amusements. They can also assess the home for hazards and provide information on the use of aids developed for the handicapped (robot smokers, non-spill cups, etc.)
- *Speech therapist assistants* can ensure daily practice and follow-up.

Any of the therapist extenders could also work in institutions to augment staff. Aides, orderlies, or other institutional support personnel could be developed using the CETA program.

These are just a few examples of services which could greatly benefit the home- or institution-bound person as well as the family. A "cascade" system of professionals and paraprofessionals could be developed to ensure adequate training and supervision. *Creative experimentation is needed in providing care for patients which can postpone long-term and expensive institutionalization.*

It wasn't skilled nursing I needed. I needed somebody to clean the house, some little things [to help me take care of my wife]... They would only allow it to be deducted if it had been a nurse. I couldn't afford a nurse and didn't need a nurse. What I needed was somebody to keep the house together.

New Health Care Personnel — Pilot Program



Genetic Counseling: More Than Genetic Facts

The Commission recommends that programs be supported to train genetic counselors, social and behavioral scientists, and other health professionals sophisticated in genetics and in the requisite counseling skills; that genetic counseling services be developed which provide access to long-term support and follow-up; and that research be conducted on the most beneficial and accessible placement of genetic counseling services, on the nature of the services to be provided, and on the impact of such services. This is to be accomplished under the auspices of P.L. 94-278, Title IV, the "National Genetic Diseases Act."

It is imperative, in a genetic disorder with late onset, terminal course, and marginal treatment, that those affected and at risk receive the most accurate information possible regarding the hereditary nature of the illness and the factors which impinge on their lives because of that inheritance. The process of conveying this information in a meaningful and compassionate fashion is called genetic counseling.

The importance of good genetic counseling cannot be overestimated. Only through knowledge can individuals, particularly those at risk, make an informed choice about major life decisions.

My son was 14 years old when we were told his father had H.D. I had never heard of the disease before that. That same evening his father took gopher poisoning, but it was too slow and too painful; he was found leaning on the horn of his car with all the doors locked. After that he was arrested for drunk driving (it was the H.D. as he didn't drink), could not hold a job, he pawned everything he owned and when that ran out, he started writing bad checks.

He signed himself to Norwalk State Hospital, but that was no help for him, so he signed himself out, rented a motel room and shot himself in the head. For 12 years now, I have lived in fear that our son will get the disease. The hardest thing to do was to tell him he is at risk. It was like pronouncing his death sentence.

Genetic counseling is a relatively new and growing discipline which has developed in response to the identification of an increasing number of diseases which are hereditary or have a hereditary component. Traditionally, counseling has been provided by Ph.D. scientists trained in genetics, or by physicians knowledgeable about genetic disorders (often the neurologist who makes the diagnosis does counseling in the case of Huntington's disease).

The Commission fully affirms the vital importance of genetic counseling to inform families regarding hereditary diseases. In the course of public testimony two important issues were raised. One regarded the suitability of traditional genetic counseling to meet the needs of Huntington's disease families. The other was the power of genetic counseling to prevent or eliminate Huntington's disease. The Commission deliberated at great length on both issues. It concluded that there must be an expanded role for genetic counseling. There must also be increased research to investigate what effects counseling actually has on people's behavior.

"Traditional" Genetic Counseling

Traditional genetic counseling is usually given in one or two sessions in which a family history is obtained and the genetic implications are explained. Information is generally restricted to the genetic facts, not legal, financial, or other matters affected by the condition. This information is often devastating. The traditional genetic counselor has no formal training in such areas as personality theory or counseling techniques and is often unequipped to help a family deal with the impact of what they are learning. As a result, the family may leave the office in a state of shock or depression. There is no follow-up to ensure that the family has a thorough understanding of the disease and its implications, and no mechanism for ongoing contact in the light of new developments. In particular, there

is no special emphasis on helping the individual or family to cope with the full meaning of the illness or to deal with the complex issues raised with regard to childbearing or other life plans the family must make.

For families struggling with a burdensome and debilitating hereditary disorder such as Huntington's disease, the traditional mode of genetic counseling will not suffice. *Long-term support and sympathetic guidance is the only treatment available for persons at risk for Huntington's disease and for the spouses of patients.* The decisions these individuals make are apt to change as they live with the disease in the family. They need access to someone who has knowledge of the disease as well as training in counseling skills.

An Expanded Definition

The vanguard of the genetic counseling profession is breaking away from the traditional mode of genetic counseling and moving toward a more comprehensive conceptualization of the process. This new definition was developed by a workshop sponsored by the National Genetics Foundation and the NIH in December 1972. With some modification, it has been adopted by the Commission's Guidance and Counseling work group:

> Genetic Counseling is a communication process that has as its goal the alleviation of human suffering associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family. The counseling tries to achieve this goal by helping the counselee to:
>
> - comprehend the medical facts, including the diagnosis, probable course of the disorder, and the available management;
> - appreciate the way heredity and environment contribute to the disorder, and the risk of recurrence in specified relatives;
> - understand the alternatives for dealing with risk of recurrence;
> - choose the option which seems appropriate to them in view of their risks and their values, and act in accordance with the decision; and
> - make an optimal adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.
>
> This definition is broad. It encompasses the long-term medical, psychological, and sociological adaptations which accompany the occurrence of a genetic disease in an individual. The extended meaning of genetic disease to the family and to society implied in this holistic definition calls for skills other than medical and genetic.

Counselors should serve as "gate openers" to a comprehensive channel of services—guiding clients from confirmatory initial diagnosis to terminal care. Increasingly, interdisciplinary teams treating persons with genetic disease are including genetic counselors in this new role.

As part of this change a new discipline of Master's level professionals is emerging—genetic associates. These individuals are familiar with the characteristics of hereditary disease. They are also trained to be sensitive to the frequent psychological complications—the guilts and depressions—which are so often a consequence of genetic disease in a family. The first genetic associate program was started at Sarah Lawrence in 1969. There are now four additional programs around the country.

Impact of Counseling on Prevention

There have been no studies to explore the impact of genetic counseling on the birth rate of persons affected by or at risk for Huntington's disease. The motivations governing individuals' decisions to have children are complex. In some individuals the wish is intense. Little is known about the equations which balance the risk of having a child affected by genetic disease against the desire for offspring. In the case of Huntington's disease the issues are compounded by many uncertainties by virtue of the disease's late onset. Much research is needed in these areas.

I love my daughters but I wish to God I had known about Huntington's disease years ago so I could have the decision to make for myself.

Although the Commission is concerned with reducing the incidence of the disease in the population, the expanded role for genetic counselors which the Commission envisions does not include prescribing the decisions Huntington's disease families should make. Genetic counseling is not a panacea for Huntington's disease. There are a number of reasons why even a vigorous program of eugenic genetic counseling would not eliminate the disease.

- It is impossible to ascertain all cases of Huntington's disease world-wide.
- New mutations will create the disease anew.
- Only totally unacceptable practices such as coercion, enforced screening, and sterilization could guarantee some reduction in the gene frequency of Huntington's disease in the population. These techniques would be applied to as many healthy individuals as those destined to become ill.

Even if the incidence of Huntington's disease were reduced, those with the illness would suffer no less. *Genetic counseling enables an individual to make an informed choice, but biomedical research is the only answer to finding a treatment and eventual cure for the disease.*

Counseling Programs under the National Genetic Diseases Act

Genetic counseling services are currently underutilized by Huntington's disease patients and families because they are perceived as only offering traditional counseling. With some modifications of existing services, they could become a valuable resource to those families and others with similar problems created by hereditary disease. *The provisions of P.L. 94-278, Title IV, the National Genetic Diseases Act,* (See p. 95) *provide the mandate for experimenting with and reorganizing genetic counseling services.*

After Mother's H.D. diagnosis, she and Daddy had consulted the parish priest about a birth control dispensation because of the hereditary factor of H.D. The priest said there were no exceptions.

The financial loss forced us into a different life style which was, and still is, difficult to accept. We stopped seeing our friends and isolated ourselves socially. Eventually we had to tell the children. How to tell them? If at that time we had had a trained social worker, or a patient service coordinator who understood our problems, the tragedy which followed would probably have been lessened to some degree. When told my oldest son became terrified and eventually became emotionally disturbed. He became convinced that he too had Huntington's disease and began to imitate my husband's behavior. He who as a youngster had been in gifted children's classes and the first few years of college was an honor student, graduated college with great difficulty and under the care of a psychiatrist. He eventually suffered a nervous breakdown and during the five years which have sinced elapsed has had seven hospitalizations, five of them in state hospitals. Although he does have Huntington's now, his fear of it has caused him tremendous suffering and has ruined the years which would have been among the most productive for him.

Coping with Emotional Stress

The Commission recommends that patients or family members who are affected by Huntington's disease and other chronic neurologic or hereditary disorders have access to affordable long-term psychological counseling and support. This should be given by trained individuals, knowledgeable about the medical aspects of the disorder and psychotherapeutically skilled, as a standard part of a comprehensive treatment plan to limit or prevent serious emotional disturbance.

The Commission recommends that Congress should appropriate funds to support pilot and demonstration projects to be initiated immediately by the Director of the National Institute of Mental Health:

A pilot program in *prevention and treatment of mental illness* in patients and family members affected by Huntington's disease and related neurologic and hereditary disorders. This program should be established with the cooperation of five Community Mental Health Centers or with center staff working in liaison with a community-based hospital or medical center.

A demonstration program to review and develop the state of the art in health psychology, beginning with a *Symposium on Psychological Response to Neurologic Conditions: Primary Prevention and Treatment.* This symposium should focus on the psychological problems of chronic neurologic illness using Huntington's disease as a prototype. Ultimately an ongoing workshop program should be maintained to develop training materials, relevant courses, research ideas, and effective communication devices.

A new discipline is beginning to emerge which concerns the relationship between physical health and psychological well-being. In contrast to psychosomatic medicine which deals with the influence of thoughts and emotions on the body, this emerging discipline looks in the other direction—the influence of the body on the mind. At innovative medical centers and hospitals around the country, health professionals are increasingly aware that life-threatening illness imposes a state of extreme psychological stress on the individual. For this reason, mental health professionals are appearing more and more frequently as members of interdisciplinary health care teams. These professionals are serving an invaluable function on cardiac units or in kidney transplant and dialysis wards, helping patients to cope with the psychological stress of acute physical illness—a stress which in itself could be life-threatening.

These same mental health professionals could serve a similar function for patients with chronic illness. Often these patients make periodic visits to their physicians, not so much for medical treatment but to relieve fear, loneliness, or boredom. Each visit is a drain on the financial resources of the individual or the Federal government, if the physician is reimbursed through Medicaid or Medicare. Mental health professionals and specially trained paraprofessionals could help these individuals cope with chronic, debilitating disease and encourage them to participate in activities which could enrich their lives. This would result in a far better use of health resources at less cost to the patient and the Federal government.

My father killed himself three months ago. He called my mother at work and told her that he could not stand it anymore, that he loved us all very much but that he could not stand to sit and be good for nothing another day. By the time she got home he had already shot himself. This was a nightmare for me and all my family.

Preventing Mental Illness

As yet there have been few experimental programs nationwide to explore the potential of mental health workers in aiding the acutely ill or chronic patient. Nor have there been studies of the role they could play in the

primary prevention of mental illness arising from medical problems. This is an area ripe for further exploration and development. It has been demonstrated that skillful interventions during periods of crisis, such as bereavement or divorce, can be a major factor in staving off serious and permanent psychological scarring.

Primary prevention programs are difficult to design because of the uncertainty of predicting who will fall ill. Certain populations are obviously in greater jeopardy than others: the poor, minority groups, those from broken homes, persons with serious (especially chronic) illness, and those from families where there is serious chronic illness.

Among the chronically ill, those with neurologic disease are highly likely to develop emotional problems. They are psychologically jeopardized by the illness itself and by their reaction to it. Neurological conditions such as Huntington's disease, Parkinsonism, stroke, multiple sclerosis, and epilepsy can give rise to personality problems as part of the disease syndrome. One aim of preventive intervention is to lessen the psychological difficulties that stem from the disorder itself. Simultaneously, the mental health worker aims to limit or prevent the additional emotional disturbance that results from the patient's reaction to the illness.

When I talk of dying personalities, I'm also speaking of my own. When you live with a spouse that's afflicted with H.D. long enough, you change too. You become irritable as you try to cope with all the problems that come along . . . Someone who cares for H.D. people needs professional guidance, too . . . We need someone who can counsel us, so we don't lose our own identities.

Family Stress

The spouse and children of a chronically ill person are also under constant stress. When the illness is hereditary, the stress is compounded. It can be expected that all members of a family in which there is Huntington's disease will develop emotional problems. Testimony at the public hearings confirmed this.

The psychological impact of the disease was considered by affected families and by professionals to be one of the most devastating problems created by the disorder. In public hearings before the Commission, witnesses spoke of psychological distress almost five times more often than any other problem.

In addition to the overall emotional impact of the disease, 62 percent of those testifying mentioned specific symptoms of social or emotional disruption which occurred in response to the disorder, either in the patient or in other family members. These included alcoholism, behavioral problems, divorce, and suicide. Over 10 percent of the oral and written testimony commented on a suicide attempt by a patient or person at risk. Half of the suicides were successful.

We've experienced medical confusion as to who can better treat Tom. The neurologists say the psychiatrists; the psychiatrists say the neurologists. We feel they both need to, because Tom's neurological disease is affecting his mental condition.

The need for ongoing supportive counseling was emphasized repeatedly in the testimony of almost 2,000 lay and professional persons and it was a major recommendation of the Behavioral Changes and Treatment work group (See Vol. II). A 1977 survey of 240 neurologists revealed that the vast majority of these specialists see their Huntington's disease patients and families only once or twice per year. However, 86 percent of those surveyed felt that families with Huntington's disease needed extensive counseling, and 69 percent felt that this counseling should come from someone other than themselves. A cogent and forceful statement of this need was given to the Commission in a position paper by Dr. Albert Cain of the University of Michigan on the "Need for Specialized Training for Clinical Psychologists":

> Whether one focuses on the psychological factors involved in patients' acceptance of or resistance to the diagnosis; the awesome realistic life-planning decisions; the frequent elements of anguish, dread, and self-loathing; the multiple cognitive dysfunctions; the tug-of-war between helpless over-dependency and false independence; the suicide attempts and completions; the marked depressive affects and clear-cut severe depressions; the antisocial behavior; the delusional, hallucinatory, or paranoid states; the psychotic or psy-

chotic-like conditions — whatever one's focus, the need for psychological understanding and services to Huntington's disease patients is evident. *Perhaps the only thing more evident is that Huntington's disease patients rarely receive such understanding or services.* (Emphasis added.)

Health Psychology

There should be a cadre of mental health workers specially trained in chronic diseases who can provide treatment for patients and consultative services for other mental health workers. This new discipline would be called "health psychology." Skills learned in working with a family with Huntington's disease are easily transferable to Parkinson's disease, epilepsy, or multiple sclerosis patients and families. For that matter, they would be transferable to any dread disease of long or short duration.

The NIMH should assume a lead role in developing health psychology as a major interdisciplinary field. This support can be channeled through programs for professional and paraprofessional training, and through mental health services. Counseling centers could be established within a medical center or in relation to a Community Mental Health Center (CMHC). Although participation would be voluntary, doctors, patients, and families would come to expect that "adjustment" or "coping" counseling, "preventive guidance," or "ventilation and education" would be a routine step in the diagnosis, referral, and treatment process. Counseling sessions should be considered as *primary prevention* of expectable emotional distress in reaction to medical problems.

Sometimes Huntington's disease patients and family members experience stress but do not seek mental health experts. They either do not recognize they are in psychological trouble or they feel that seeing a mental health professional means that they are mentally, as well as physically, ill. For many, mental illness is still a stigma or a sign of impending disease. *Routine counseling as a standard adjunct to good medical care can prevent the serious emotional consequences of physical illness—before the damage is done.* The Commission urges that programs to explore ways to provide supportive psychological counseling and to develop the field of health psychology be initiated at once.

The commission recommends that Congress appropriate funds to support two pilot and demonstration projects to be initiated immediately by the Director of the NIMH.

A pilot project in prevention and treatment of mental illness in patients and family members affected by Huntington's disease and related neurologic and hereditary disorders.

Psychological counseling programs should be developed in cooperation with five CMHCs or with center staff working in liaison with a community-based hospital or medical center. The purpose of these test programs would be to determine the best way to provide psychological services adapted to the needs of Huntington's disease patients, those at risk, other family members, and those with related disorders.

CMHCs have the potential to serve the needs of patients and families with chronic neurologic and hereditary diseases. However, some medical knowledge of these diseases and their impact is necessary for effective counseling. Most professional personnel at CMHCs are not trained to recognize these disorders. They do have valuable skills in family counseling, individual and group psychotherapy, and crisis intervention. With only moderate additional medical training, consultation with experts on these diseases, and some experimentation on the most efficacious ways to organize and deliver services, the CMHCs could become a highly valuable component in a comprehensive community support plan for families with Huntington's disease and related disorders. The Staff College of NIMH could be used in education and training.

A demonstration program to review and develop the state of the art in health psychology, beginning with a Symposium on Psychological Response to Neurologic Conditions: Primary Prevention and Treatment.

As an initial step in consolidating and developing the field of health psychology, a symposium should be held focusing on the psychological problems of chronic neurologic illness. Huntington's disease should serve as a prototype. The symposium should be relatively small and consist of invited speakers. The proceedings should be published as a contribution to the literature in the field. Ultimately, an ongoing workshop program should be maintained to facilitate communication, develop training materials, and stimulate research ideas.

A Right To Work

The Commission recommends that the Secretary of the Department of Labor (DOL) ensure that all persons with Huntington's disease and other neurological or otherwise handicapping conditions have the benefit of work suited to their capabilities, from sheltered workshops to full employment.

Guidelines must be developed for physicians who evaluate patients and make job placement recommendations. The support of concerned health voluntary organizations should be solicited in hiring the handicapped.

The Secretary should also guarantee the rights of Huntington's disease patients, those at risk, and others who are either handicapped or perceived to be handicapped, to affirmative action and antidiscrimination protections through stringent enforcement and vigorous outreach as required by Section 503 of the Rehabilitation Act of 1973.

An increasing number of diseases are being recognized as genetic in origin or as having a genetic component. This has raised a number of intriguing and complex issues in employment. The three categories of individuals who are of most concern are (1) those who are carriers of a recessive gene, but who will not be affected by the disease (e.g. sickle cell carriers); (2) those who have a genetic disease and who may be handicapped or impaired by it (e.g. a patient with Huntington's disease or sickle cell disease); and (3) those who are at risk for a dominant disease who have a 50 percent chance of developing the illness. Persons in all three categories are now in the labor pool.

Job Discrimination

Healthy carriers of a recessive disease trait have been discriminated against by employers who do not understand the nature of genetic diseases. The recent well-documented story of discrimination against carriers of the sickle cell trait is evidence that a little learning about genetics can be a dangerous thing.

People who develop a genetic illness must be evaluated individually to determine their capabilities, as are people with non-hereditary disabilities.

Individuals who are at risk for an inherited disorder pose special problems, epitomized by Huntington's disease: there is no way of detecting who will develop Huntington's disease before symptoms appear. One-half of those at risk will develop the disorder; the other half will not. Before the disease develops, both groups are presumably normal. But since onset of the disease occurs in the thirties or forties, career commitments have usually been made.

My youngest nephew has had his goal set on being a doctor all of his life and has worked toward that aim. He is an excellent student and has worked for the finances involved in such an education. He is now faced with problems in being accepted in medical school because he is a member of an H.D. family. He is attempting to lead as normal a life as possible but society will not allow it. He is being made to feel "guilty" about having a father who died of H.D. when the committee who interviewed him for entrance into medical school admitted that the reason for his not being accepted was H.D. Such barriers cannot be broken down in a short time but some measures need to be instituted to educate people, even people associated with the medical field.

Legal Protections

Section 503 of the Rehabilitation Act of 1973 ensures strong protections for the handicapped against discrimination in hiring practices by employers receiving even minimal amounts of Federal funds. Those who are carriers of recessive disease traits or at risk for genetic diseases but healthy, are protected under one of the definitions of handicapped developed for the implementation of

Section 503—those who may be *perceived* as handicapped. Those with diseases, genetic or otherwise, are protected under another definition—those with handicapping conditions.

The Commission is seriously concerned that the general public, including employers and employees, may not be aware of these protections against employment discrimination. In testimony to the Commission 7.5 percent expressed the wish to remain anonymous, primarily because they were afraid of losing their jobs or of jeopardizing insurance policies. There is a need to educate employers, the labor force, the general public, and health voluntary organizations about the nature and extent of Federal protections against employment discrimination under Section 503. There is an additionally pressing need to educate people concerning hereditary disease. As screening for genetic conditions such as sickle cell anemia, Tay-Sachs disease, and thalassemia become more commonplace and new screening tests for other heritable disorders are developed, there must be vigorous efforts on the part of the DHEW, the DOL, and the health voluntary organizations to ensure that those affected by hereditary conditions do not become targets for discrimination.

The DOL should actively solicit the aid of health voluntary organizations in helping employers meet their required quotas of handicapped employees. The Huntington's disease health voluntary groups could serve as intermediaries between employers seeking job applicants to fulfill affirmative action requirements and capable Huntington's disease patients looking for work.

Not only must employers take affirmative action to hire the handicapped, but the DOL must give affirmative leadership in ensuring that all those disabled who are capable of working can find work.

In so doing, the DOL should also support and increase cooperative government and industry employment projects, including sheltered workshops similar to the current Training and Placement Service Programs (TAPS) funded by the CETA.

Our son had an ROTC scholarship at Colby and was to be a pilot in the USAF; after he finished basic and went to flight school some doctor discovered on his medical record his father had H.D. He was immediately grounded and treated as if he had nothing more to live for. Finally after months of telling him they were going to discharge him they changed their minds and sent him to radar school instead.

A newspaper reporter called me to inquire if I would come downtown and talk with a man who frequented the paper only at night. He was wearing a silver bracelet that said, "I suffer from Huntington's Disease. I am not drunk."

It was 3:00 a.m. in Chicago. I met a tall, thin man with blonde curly hair and beard. He looked to be thirty years old. He wore overalls and three flannel shirts, one on top of the other. He carried a torn blanket held in a ball-like fashion under his arm, freeing his hands to touch walls, doors, or stairs as he walked the jerky, irregular wide gait of a Huntington's person.

It turns out he had been a pilot in the war overseas. He could go to any veterans hospital for care but said, "Not yet." "Why," I asked. "They don't let you exercise enough. I got weak there so I left." "When?" "Oh, not long ago."

Gradually he told me about being a pilot—some memories on his mind about the war; earlier about school. Basketball. Before that, a divorce of his parents. The illness of his mother. She died at home; she had Huntington's. He touched his I.D. bracelet, too. Who took care of her? He said, "I did." His body bent forward. Tears.

"Who is looking out for you?" He answered firmly, "I am."

"Where is your father?"

"I don't know." He looked away. "We're out of touch, my father and I," he said.

"How do you manage?"

"I get a disability check but it's only $99.00. I can't live off that." He held out his left hand and the other was moving in the air, jerkily. "So I have it sent to a hotel. I pick up the check, cash it at a currency and live on the streets." He took showers at Mayor Daley's gym on Navy Pier, ate in a restaurant, and when his money ran out, he asked churches and synagogues for money. "They have never let me down yet." He said he washed his inner shirt and put it on the outside to dry as he walked around the city. He slept on the beach and went to The Salvation Army to sleep on a cot when it rained.

Some doctor gave him a silver bracelet that helped him the most, he said, because he could show it to the police if stopped. This usually made quite a difference in how he was treated. Before he got the bracelet, the police would rough him up and treat him like a drunk. He frowned and said, "I've been beaten up on the streets."

I asked him if he knew where the Veterans Hospital was near the beach where he slept. "No," he said. "Would you like me to show you where it is?" "Yes."

When we got to the V.A. Hospital and Rehab. Center, it was locked up. No guard at the door. I was looking for an emergency room. It was closed, too.

We stood there in the light from the hospital basement and the man said, "You want me to go in there, don't you?" "Yes, I do but it's up to you." He said, "It's not time yet."

He had worked out a precious routine for himself; maybe he was right. There will be time for hospitals. "Thanks. I needed to talk to somebody," and he extended his hand. I watched him walking swiftly, arms and legs moving freely until he was well out of sight. I never saw him again.

Rehabilitation: Enriching Lives

The Commission recommends that the Commissioner of the Rehabilitation Services Administration (RSA) designate persons with Huntington's disease and related disorders as underserved groups. The initiation and expansion of services to this group should include the training and orientation of rehabilitation counselors regarding Huntington's disease and related disorders. Adequate psychological counseling and assistance in job-finding, or adjusting to living at home or in the community at a maximal level of functioning should be provided.

The Rehabilitation Act of 1973 should provide ways to enable handicapped individuals to find employment. It should also assist those who cannot be expected to work to live as independently as possible with their families and in the community. The emphasis in the Act should be on the *overall* rehabilitation of handicapped individuals, not solely *vocational* rehabilitation.

People with Huntington's disease, and many others with progressive illness, have been turned away from rehabilitation programs. According to a narrow interpretation of the law, rehabilitation counselors can refuse to provide services to these individuals. They interpret the law to mean that handicapped individuals should be brought to some stabilized level of functioning which enables them to seek and sustain work. Huntington's disease patients have progressive and terminal disease—their work future is limited.

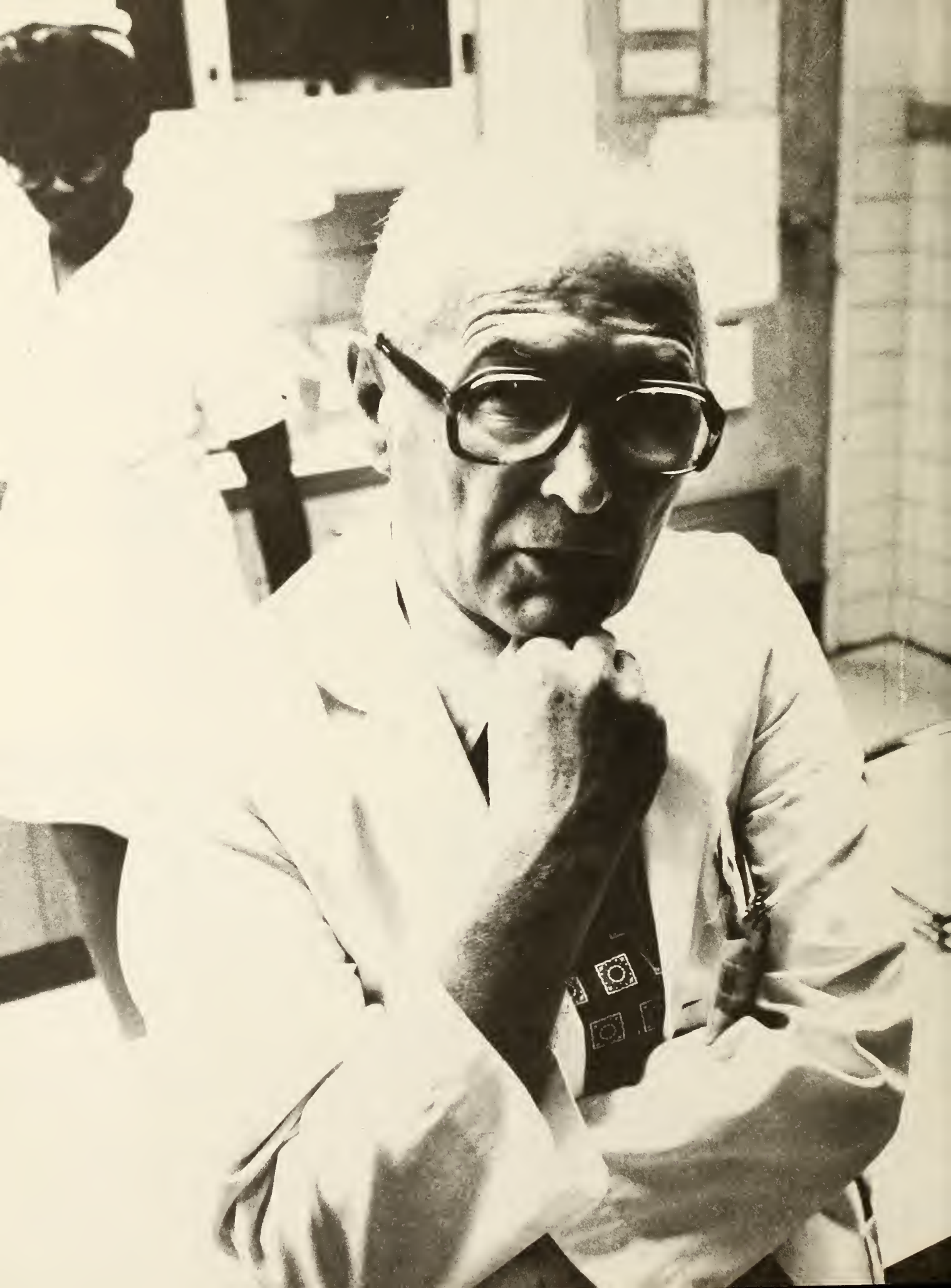
He's dependent upon others, mainly his immediate family. He's bored stiff most of the time. He cries a lot. He feels he has nothing left to live for. He's only 36!

Yet Huntington's disease patients may have 10 or more years in which they could be engaged in constructive activity, not whiling away their hours in boredom and depression.

The diagnosis was confirmed at age 24. Since that time he has been employed at the North Central Washington Supervised Skills Workshop. It has been a wonderful answer to the problem of keeping him occupied with something besides T.V.

Many are forced to retire early from jobs requiring fine coordination or intense intellectual demands. Although a Huntington's disease patient may no longer be able to practice medicine or fly an airplane, there is much that such a patient could do, either for remuneration or otherwise. When patients can no longer work, they should be counseled in adjusting to living with their families or in the community as well as they can.

Applicants for rehabilitation services should be assessed individually, whether they have a progressive and terminal illness or a stabilized condition. Rehabilitation services should be available to individuals throughout the course of a long and progressive illness, particularly if there are plateaus of stabilization in the process. Services should be tailored to the stage of the illness.



Misinformation: Mandate for Education

The Commission recommends that Congress appropriate funds for the development and dissemination of educational materials on Huntington's disease for families, health care professionals, community service personnel, and the public.

A national information clearinghouse should be established for the purpose of developing, collecting, and distributing educational materials on human genetics, genetic disorders, and birth defects, as a means of implementing P.L. 94-278, Title IV, the National Genetic Diseases Act.

Problems of misinformation, mishandling, and misunderstanding plague families affected by Huntington's disease and other hereditary disorders. Surprisingly, the problems often begin in the doctor's office or in encounters with other health professionals. In public testimony to the Commission, patients and families described the need for *professional education* as one of the most urgent recommendations. Public education and family education were also rated among the highest priority recommendations.

Misdiagnosis

Misdiagnosis is one of the most damaging consequences of a lack of professional education regarding Huntington's disease. This was reported as being among the most serious difficulties individuals encountered. The most common misdiagnoses were for schizophrenia and Parkinsonism, but the list included even polio of the throat. Exorbitant sums were spent in useless and frustrating rounds of doctors before an accurate diagnosis was achieved.

After hospitalization and seeing 31 doctors for services totaling over $36,000, one doctor recognized the disease.

There are problems in diagnosing hereditary diseases which affect only small populations of patients. Some doctors may never have seen a patient with Huntington's disease or may have long forgotten seeing such a patient while in medical school. But the problem of unfamiliarity with hereditary diseases is compounded by current practices in medicine and medical education:

- Doctors seldom see families over successive generations and so are not likely to be aware of a hereditary disease pattern.
- Human genetics is taught in the early years of medical school and is often not integrated with subsequent training in individual diseases.
- Mental health professionals are often the first medical specialists consulted by a patient with Huntington's disease; this group was frequently indicted by witnesses because of their failure to diagnose the disease accurately. Presumably these specialists are not sufficiently aware of the necessity to look beyond psychiatric symptoms to seek a possible physical cause.

Misinformation on Inheritance

Even when an accurate diagnosis of Huntington's disease was made, many patients were given misleading information on the inheritance pattern *by the very professionals who diagnosed the illness.* Acting on this misinformation, some couples decided to have children. When they were later told that they might have inadvertently passed on the defective gene, they could only feel helpless rage and despair.

Inaccurate diagnosis of Huntington's disease and the consequent lack of genetic counseling deprives individuals of the only opportunity they have to choose not to pass on the gene. *If patients and families are not properly informed, they have no choice.*

Mishandling of Nursing Care

Many of the nurses and allied personnel who testified before the Commission complained of a lack of available information on how to handle the often complex problems of care presented by a Huntington's disease patient. Nurses must use trial and error methods, and there is no mechanism by which techniques learned in practice can be broadly disseminated.

Neurological nursing care is a nursing specialty demanding special techniques. It requires compassion and skill in handling persons with movement and coordination problems, intellectual impairments, and difficulties in talking or swallowing. There are few special training programs for nurses in this area, and no neurologic specialties for nurse practitioners.

Unlike some of the more popular areas of nursing practice, neurologic conditions—which are poorly understood and not readily amenable to medical management—are often shunned by nurses. If such conditions receive the focus of federal, state and private agencies (as do cancer, stroke and heart problems at this time) there would be more impetus for nurses to go into this area of practice. When such conditions are seen in the light of what they really are and people respond accordingly, neurologic medicine will move from the dark ages to enlightened, enthusiastic patient care.

Misunderstanding Stress

Any serious disease affects the whole family as well as the sick individual. A hereditary disease increases the burden on all—on the patient who may have passed on the disease, on the spouse, on relatives who are at risk, and on their spouses and children. Because of its psychiatric symptoms, Huntington's disease can further disrupt the family. Physicians, mental health professionals, nurses, community service personnel, and the general public must be made aware of the stresses hereditary disease can impose. Not long ago a Detroit police matron scolded a 12-year-old girl at risk for Huntington's disease: "You'd better behave or you will get sick like your mother!" These horrors must stop.

While we were there her doctor casually mentioned, "Oh, you did know that she has Huntington's." My dad and I were stunned. They knew this for two years but somehow never bothered to tell us. I was very irate because I'm 24; I could have had a child by now, and I feel like they had a responsibility to inform me. If I would have had a child, I would be outraged.

Misapprehension and Fear

Many families know nothing about Huntington's disease until it is diagnosed in a family member. They have no idea what to expect or how to care for the patient. The physician attending the patient may have had little or no experience either. Both family and physician need help in knowing what to anticipate as the disease progresses, how to prepare the patient and at-risk family members, and how to provide proper care. Almost no written or visual materials exist to provide this information.

Educational Goals:

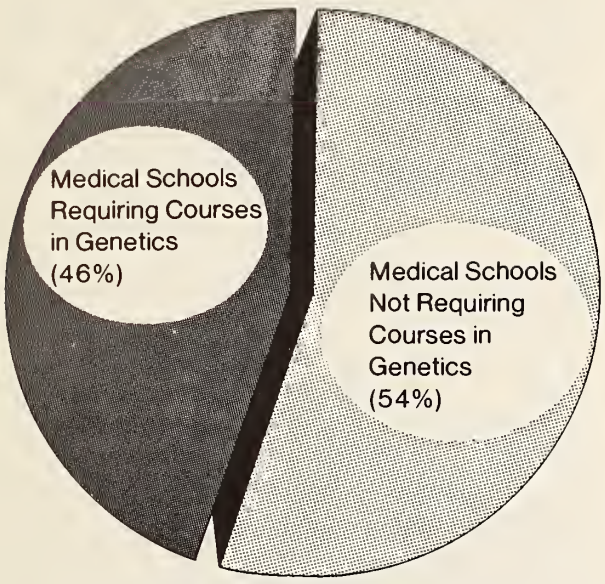
Patient and Family Education

Huntington's disease patients and their families must have the most thorough and up-to-date information on the disease and rapid access to new information as it develops. Materials should include a description of the illness and its course, as well as details of care and information regarding medical, social, and financial assistance. This information should also include simplified details on new developments that are taking place in both research and treatment. *Educational materials should be developed with the aim of enabling patients and families to take an active part in their own care.*

Health Care Professionals

The traditional approach to teaching medicine disease-by-disease was tolerable when there were few genetic diseases recognized. Today there are approximately 2,300 disorders known to be inherited, each one affecting a small population of people. Rather than trying to memorize individual disease entities, physicians must learn to recognize symptoms common to a variety of genetic disorders and include inherited diseases in their differential

Medical Schools in the United States Requiring Courses in Genetics



Total Number of Medical Schools: 120

Source: *American Association of Medical Colleges 1976 — 1977 Curriculum Directory.*

diagnoses. Physicians must become more conscious of the possibility of genetic factors at work in the illnesses they treat, including those which may have some hereditary component such as cancer, heart disease, diabetes, and schizophrenia. Many untreatable debilitating or lethal genetic diseases could be prevented if individuals knew of the disease's hereditary nature before they had children. Other health care professionals should also be trained and have access to information on treating Huntington's disease patients and families.

Service Agencies

These groups include the Social Security Administration (SSA), VA, Health Care Financing Administration (HCFA), DHEW, employers and employment agencies, insurance companies, legal and justice systems, advocacy and volunteer groups, social service agencies, clergy and church groups, legislators, and vocational rehabilitation agencies. These groups do not have to be medical experts, *but they do have to know where they can obtain accurate information on Huntington's disease and related disorders when the need arises.*

General Public

Almost every family has a notion of "something that runs in the family," but the general public is just beginning to become aware of genetic diseases or predispositions to genetic disorders. Education about human genetic diseases only entered high school curricula in the 1950s. Even today, human genetics is given short shrift in most secondary school and college biology texts, where the emphasis is usually on the genetics of rats and fruit flies.

Patients and families with Huntington's disease feel ostracized and stigmatized in a society which is unfamiliar with the illness. Studies indicate that people who are only carriers and not afflicted by hereditary illnesses, such as those with Tay-Sachs or sickle cell trait, often feel ashamed and conceal their status.

I have become acutely aware of how little I know about H.D., both as a person and as a physician. I have looked through my medical textbooks, and am appalled at the paucity of information I can find about the patient with H.D., as opposed to the pathology specimen.

Genetic education is preventive medicine. The goal for the future is to develop educated citizens who are aware of the variability of human genetic nature. Individuals should know about their own genetic inheritance so that they can take appropriate steps to prevent illness, such as dietary or occupational precautions. All should be familiar with the ethical and social problems associated with human genetics and genetic disorders.

Educational Materials

In order to implement the Commission's recommendation for the development and dissemination of educational materials and programs on Huntington's disease, the following actions should be taken:

- The creation and distribution of educational films, brochures, exhibits, curriculum additions, magazine articles, radio/TV programs, or any other appropriate educational vehicles to explain Huntington's disease to the target audience—

whether it be the Huntington's disease patient and family, specialized health care professionals, community service agencies, or the general public.

- The development of educational materials, particularly for nurses and other personnel (including family members) providing direct care to Huntington's disease patients and others with similar neurologic disease. These would include videocassettes demonstrating effective patient/family management of daily living in the home or in institutions. Such materials should meet the American Nurses' Association standards for continuing education credit.

- The development of courses in human genetic disorders and their integration into medical school and continuing medical education curricula. These materials should be prepared jointly with the American Medical Association and specialty groups in cooperation with medical schools throughout the nation.

A National Genetics Clearinghouse

Congress recognized the need for education and informational materials on genetic diseases when it passed P.L. 94-278, Title IV, the National Genetic Diseases Act. (See p.95.) The Commission believes that a national clearinghouse for the development and dissemination of materials on human genetics, genetic disorders, and birth defects would facilitate access to information. A nationwide and coordinated pool of educational materials would best realize the intent of P.L. 94-278, Title IV, the National Genetic Diseases Act.

This clearinghouse should be created either within an appropriate governmental agency or by contract to a private institution. The clearinghouse should be responsible for creating and acquiring textbooks, teaching aids, brochures, pamphlets, guide books and manuals, films, and workshop training materials to be used in training and educational activities. It should also serve an outreach function through workshops, speakers' bureaus, etc., in reaching target audiences who could benefit from an increased awareness of human genetic diseases.

When I arrived at the hospital, the very young doctor there felt I needed to be instructed—which I did—in all of the ramifications of Huntington's disease. And he gave me the whole load in 20 minutes. And the first thing he told me was, do not have any children. And I was at a loss to say, "What do I do with the children I already have?"

I realize, he was giving me the proper information, but somehow he had forgotten that I had feelings, and that the trauma was almost more than I could assimilate at that one moment. And so, I really felt that, somehow, we needed to educate, not only the brain, but the heart for when they have to talk with people who must face this Huntington's disease.

He had been told by members of a hospital board in Dayton, Ohio, where his mother was, that the chances of him or his offspring getting H.D. were one in a thousand! This was in 1961.

My daughter was born at Emory University in Atlanta—a very fine place. When I went to this obstetric group there, my mother-in-law was always in the back of my mind, and I said, my mother-in-law had Huntington's. Oh well, that's all right, nothing to worry about! Each obstetrician that I had—God is my witness—I brought this up, and each one said, don't worry about that! Oh I would never have had these children . . . The doctors need education; we need education.

The doctor told my mother when her mother died, that it was none of her business what she died of.

It was only through an autopsy performed on her brother that we were finally able to determine what caused the horribly weird behavior and deterioration of a once beautiful person. Prior to the autopsy, it was years of tests, hospitalization, psychiatrists, mental institution, shock treatments, etc. for her. Had we known what was wrong with my mother, we could have avoided so many tragedies which are too numerous to name.

One of our doctors here says it's so rare no one has it—another says, "Oh, that's no problem now. They treat it with L-Dopa.

From 1944 to 1955 a succession of doctors and several clinics tested and treated my mother for female problems to tumors before being sent to the Mayo Clinic for the correct diagnosis.

The opinion of the townspeople was that he was a drunk. He was even jailed because of his uncoordinated walk and slurred speech.

The Promise of the National Genetic Diseases Act

The Commission recommends that P.L. 94-278, Title IV, the National Sickle Cell Anemia, Cooley's Anemia, Tay-Sachs and Genetic Diseases Act, be extended and that the full authorization of $30 million be appropriated annually for FY 1979, FY 1980, and FY 1981.

The Commission recommends that an administrative unit be established in the Office of the Assistant Secretary for Health which would be responsible for planning, evaluating, and coordinating genetic services and research, monitoring grant review and allocation of funds, establishing program and funding priorities and planning policy with respect to the social, economic, legal, ethical, and psychological problems that are anticipated with the development of new technologies in genetic screening. Input from non-government professionals and consumers should be solicited. This office should have responsibility for monitoring the implementation of those sections of the Huntington's Disease Commission National Plan which have relevance to the provisions of the Act.

In 1976, Congress passed the National Sickle Cell Anemia, Cooley's Anemia, Tay-Sachs and Genetic Diseases Act (P.L. 94-278, Title IV). The purpose of the Act was to establish a national program of "basic and applied research, research training, testing, counseling, and information and education programs with respect to genetic diseases." Thirty million dollars was authorized annually for FY 1976, FY 1977, and FY 1978 to support new initiatives in screening, counseling, information, and education. No new monies were appropriated for the Act in FY 1976 and FY 1977. The Labor-HEW appropriations bill for FY 1978 for the first time includes a $4 million appropriation for Title IV, only 13 percent of the original authorization.

PHS Genetics Coordinating Committee

In June 1977, the PHS Genetics Coordinating Committee was formally chartered to serve as a forum for discussion and a focal point for the coordination of PHS activities relevant to the provisions of the Act. Committee membership includes representatives from all bureaus, institutes, and divisions of the NIH and all PHS agencies with programs involving genetic disease research or services. The Genetics Coordinating Committee is to advise the Assistant Secretary for Health and PHS agency heads on the status and technical adequacy of PHS activities with regard to genetic disease.

There has been little policy planning or establishment of priorities with respect to the National Genetic Diseases Act, although it was passed in 1976. No leadership has emerged from the Health Services Administration (HSA), the agency designated to plan and carry out implementation of major provisions of the Act. The first annual report on the administration of the Act, submitted to Congress in compliance with the reporting requirements of the Act, merely describes existing programs on genetics within the PHS. No evaluation of these programs or estimates of future needs was included.

Eventually she was referred to a specialist who made the H.D. diagnosis. I will never forget his saying, "This disease is inherited but it only appears every third generation, so you and your brother and sister don't need to worry—by the time the third generation is here, they will have found a cure!" Never having heard of H.D. we did not question this statement . . . They say ignorance is bliss—we thought we were safe and H.D. went out of our minds. In the ensuing years, my sister and brother and I had children of our own.

Genetics and Public Health

The Commission finds these events disturbing in view of the overwhelming impact of genetic disorders on the public health and on the well-being and integrity of the family. *Inherited diseases are, above all, family diseases.* They have profound effects on all members of the family, whether they are possessors of the genetic trait in question or not.

Because of changing disease patterns and new knowledge regarding the pathogenesis of disease, a growing proportion of the nation's health problems are now recognized as genetic. An estimated 6 percent of the population, over 12 million people, suffer from severe genetic disease; the figure increases dramatically when multifactorial genetic conditions such as diabetes and schizophrenia are included. The economic cost to the nation of caring for those afflicted with genetic diseases amounts to billions of dollars annually. The cost in family stress and individual suffering is incalculable.

The importance of genetics was emphasized in the President's Biomedical Research Panel:

> . . . The ultimate prevention of psychoses, diabetes, high blood pressure, coronary heart disease, and lung cancer becomes more likely with an understanding of the degree to which genetic factors underlie susceptibility to these diseases. Appropriate preventive schemes can be directed to the small proportion of the population which is at high risk.

For these reasons, the care with which the National Genetic Diseases Act is implemented is of utmost importance. There is a crucial need for thoughtful planning and evaluation of new and existing programs. Although nine diseases, including Huntington's disease, are specifically mentioned in the Act, the law requires new, noncategorical initiatives in genetic research and services. For the first time, hundreds of separate categorical interests can be brought together under one act. Rational and cost-effective programs can be developed based on meeting common needs rather than satisfying separate specialized concerns.

Flexible Funding

Much of the activity specified in the Act will involve the state and local health departments, but allocation of Federal funds to these organizations should be only one mechanism by which programs are implemented. Alternative mechanisms allowing for the funding of selected demonstration programs of novelty and merit should be sought. Funding of programs must be as flexible as possible. An equitable allocation of funds among agencies must be ensured and some mechanism devised to transfer funds easily as the need arises. New research on the delivery and impact of genetic services must be encouraged and supported. All of these issues have yet to be addressed in any implementation plan.

The Need for Thoughtful Planning

The Commission believes that it is vital to establish a new organization to plan policy, evaluate programs, and advise on a wide range of interdisciplinary issues involving research, detection, prevention, and treatment of genetic diseases.

The Commission firmly contends that in the next decade there will be a burgeoning of knowledge about inherited diseases. Techniques will be developed to screen for a host of crippling, debilitating, and lethal diseases. It is hoped that treatment methods will keep pace with innovations in detection and prevention. These discoveries will create problems as well as solutions. There will be agony for individuals who learn they are destined to develop a disease for which there is no treatment. There will be profound ethical, legal, and psychological problems for families who learn that their unborn child is affected by a genetic disorder. New strains will be placed on the confidentiality of the doctor-patient relationship. Problems and complex issues will be raised in employment, in obtaining medical and life insurance coverage, and in qualifying for professional training programs.

Enough information is now available to anticipate these problems and plan wisely. It is irresponsible to do nothing but wait until technology overtakes society. The Commission urges that an administrative unit be formed to address these issues immediately.

An OASH Unit

The Commission considered a number of alternatives for the organizational structure of this unit. One possibility would be an Advisory Panel. But the Government has seen a plethora of advisory panels, and while many serve a useful purpose, they can also impede progress by requesting innumerable reports and otherwise interfering with day-to-day activities of the agencies they advise. Panels do, however, provide visibility and focus to a problem which can otherwise be obscured.

An office within the NIH or the HSA was another possibility considered. The major impediment to this plan is that genetic

research and services extend over so many agencies, both inside and outside the PHS, that an office in any one institute or agency might not have sufficient authority to oversee all agencies in monitoring the Genetic Diseases Act.

A third option is to create a new office at the level of the Assistant Secretary for Health. This idea has great appeal because at that level of authority the office would be in a better position to coordinate programs involving individual agencies of the PHS. An office at the Office of the Assistant Secretary for Health level would also have greater flexibility and authority in establishing contacts outside the PHS, for example, with the DOL, the VA, the Office of Science and Technology, and the Domestic Council.

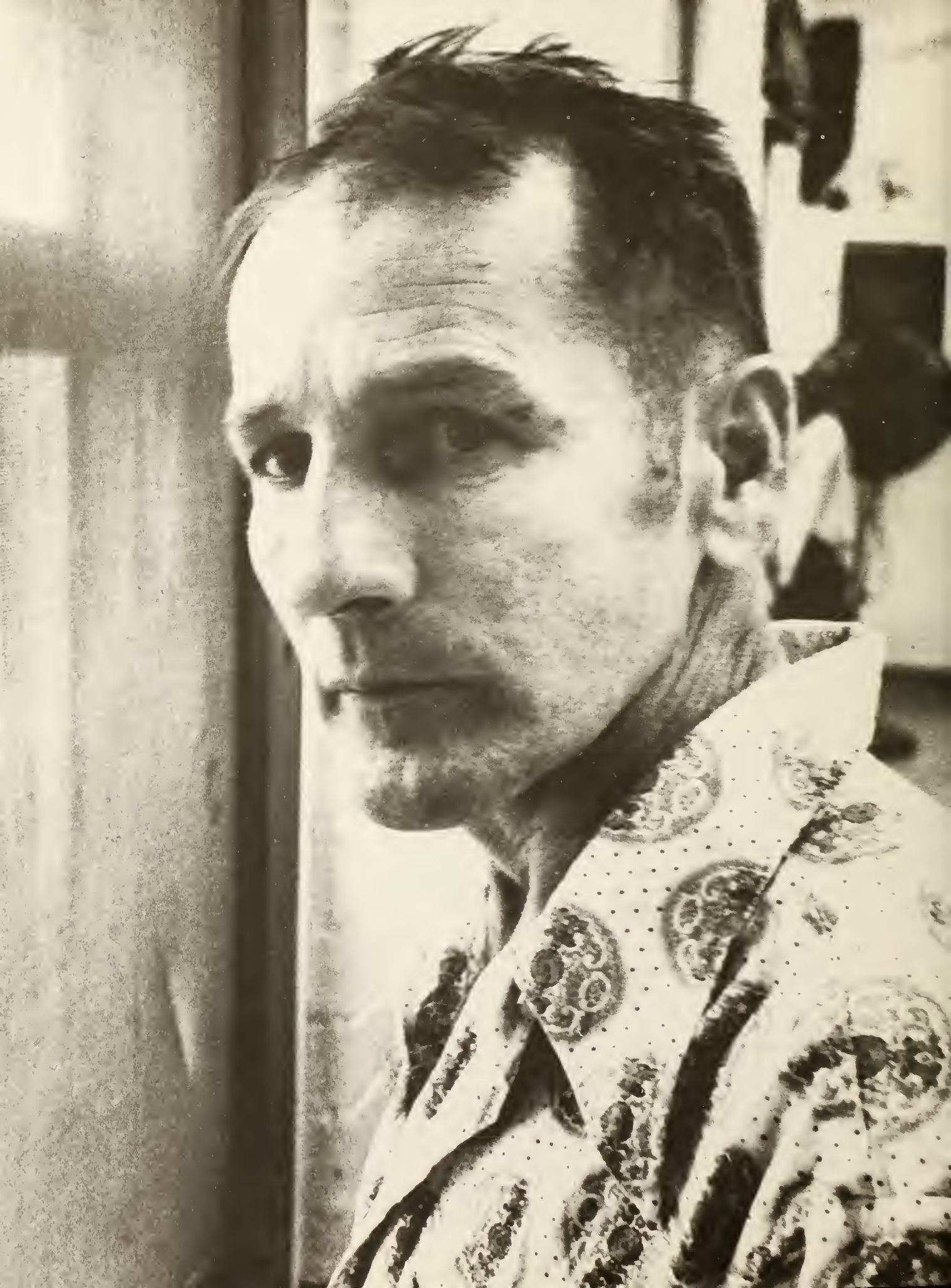
The Commission prefers the last option of those considered. Such an office would provide visibility as well as the capability of broad policy planning across the PHS.

The Commission therefore recommends that the functions of policy planning, evaluation, and coordination be carried out by an identifiable administrative unit within the Office of the Assistant Secretary for Health. This office should review grants and allocate funds or advise and oversee any other office which has this responsibility. Non-governmental consumer and professional expertise should be sought in developing any implementation plan and in continuing policy planning and evaluation in genetic services and research.

Oversight Responsibility

The National Plan developed by the Commission is a comprehensive plan designed to meet the research and service needs of Huntington's disease and other hereditary disorders. As such, the implementation of relevant sections of the National Plan should be included in the implementation plans developed for the National Genetic Diseases Act. The administrative unit planning policy and coordination for the Act should also have oversight responsibility for relevant sections of the Huntington's Disease National Plan. The annual report to Congress required by the Act should describe progress in the implementation of the Plan.

My husband is the one that's afflicted with it. I didn't know about it. It must have been in his family for a long time, which they didn't really recognize. His mother died at the age of 36, and her mother died earlier . . . It was just like a completely different person than what we were used to. He'd beat me, he'd beat the children, he'd break things. He'd lose jobs, and this would cause bad feelings, because any time that you've got six children, you've got to support these children. And then he lost job after job after job . . . He's been in the Veterans Hospital for about ten years . . I visited him the first year or two after he was out there and I took the three older boys with me. And it shook them so bad to see their father in the condition that he was in, they knew that there was something wrong with him, because of the way he treated them. They knew that he wasn't like other dads, you know, because they had visited with other children and they knew he was different . . . And just two years ago they asked me for consent to put a tube into his stomach. They had been feeding him—I guess they call it through the nose—and he would pull this out. And so, they asked me about putting this tube into his stomach. Well, that's what they did then. And three years ago is when we took Russell to the Kansas Neurological Institution and they were good people . . . He was 14. The thing of it was, his was different because his came on him—like, he was only six years old when I first started to notice this. And then, you know, he was a big boy, six feet tall and he got to the point where I wasn't able to really handle him with the children that I had at home, plus I was having to work out and be the financial support of the family . . . And so, I put him down there . . . And he died this January . . . And the main thing of it is, it's not only him. I've got a boy at home that will be 14 in August that's got the very symptoms of this terrible thing. And the doctor diagnosed this. And so, I just say that I've spent 20 years of my life that's been nothing but tragedy.



Implementing the National Plan

The Commission recommends that a branch for Huntington's disease and related disorders be created within the Neurological Disorders Program of the NINCDS. This branch would be charged with overseeing the implementation of the National Plan for the Control of Huntington's Disease and Its Consequences. The branch should be represented on the Public Health Service Genetics Coordinating Committee. It should coordinate activities with the proposed Genetic Services and Research Unit in the Office of the Assistant Secretary for Health.

The National Plan developed by the Commission was formulated to meet the needs of Huntington's disease patients and families. In the process, the Commission has sought solutions which can go beyond Huntington's disease to effect changes in broader health problems in America today. Its implementation will ease the plight of Huntington's disease families and lead toward the promotion of better health for all Americans.

Some of the Commission's recommendations can be initiated immediately at the NIH, following Congressional authorization and appropriations. These recommendations include increased research in genetics and the neurosciences, the establishment of a National Tissue Bank, Centers Without Walls, and such programs as the Venezuela project and interdisciplinary workshops.

Other recommendations, e.g., pilot programs of Health Services Coordination and Development or implementation of provisions of the National Genetic Diseases Act, involve agencies outside NIH but within the PHS. Still others require planning, research, and coordination across major agencies and departments of the Federal government. These include coverage of long-term care of the chronically ill in any program of National Health Insurance, establishing a Task Force to find ways of developing drugs of limited market value, and creating new health care jobs at both professional and nonprofessional levels.

All recommendations can and should be implemented at once. All will require careful follow-up by someone with authority to ensure that the full intent of the Plan is carried out and that individual provisions are acted on promptly and thoroughly. Because some recommendations can be implemented within a single agency while others require interdepartmental planning, the Commission proposes two levels of implementing authority: one would be within an institute of the NIH; the other would be at the level of the Office of the Assistant Secretary for Health.

A New Branch of NINCDS

The Commission feels strongly that the lead institute for neurology at the NIH—NINCDS—should be the primary overseer of its research and pilot project recommendations. The Commission proposes that a new branch for Huntington's disease and related disorders be created within the Neurological Disorders Program of the NINCDS to supplement the two existing branches: Epilepsy and Developmental Neurology.

The heightened visibility such a branch would create would in itself recruit interest in research or clinical studies of Huntington's disease and related disorders. It might also serve to unify the interests of the many separate health voluntary organizations concerned with movement disorders or other related neurological diseases who currently look to NINCDS as their principal source of hope.

Liaison with OASH Unit

Because Huntington's disease is a hereditary disorder as well as a neurological disease it is important that there be close liaison between the new branch and the proposed administrative unit within the Office of the Assistant Secretary for Health. The Commission has recommended that such a unit be established to coordinate genetic services and research throughout the PHS. In particular, the Commission recommends that the Office of the Assistant Secretary for Health unit have oversight responsibility for those sections of the Huntington's Disease National Plan which are relevant to provisions of the National Genetic Diseases Act, P.L. 94-278. To facilitate liaison with this office the Commission recommends that a representative of the new

NINCDS branch for Huntington's disease and related disorders be included in the PHS Genetics Coordinating Committee, and work closely with the Office of the Assistant Secretary for Health unit on sections of the Huntington's disease plan relevant to genetics.

Both the NINCDS branch and the proposed Office of the Assistant Secretary for Health unit should be responsive to the Huntington's disease health voluntary organizations or their designated representatives in providing follow-up information, progress reports, or other data relevant to the execution of the National Plan.

Perhaps it appears that I expect too much. I realize that there are many sick people in our country who do not receive good medical care. I know that there is nothing that can be done about the lack of consideration in people. I also realize that governmental agencies are hampered by bureaucracy . . . However, considering the hereditary aspects of the illness and the extreme disability it causes, I feel that extreme means are necessary to do something about it.

In spite of the social, financial, and psychological impact of Huntington's disease on the family, I feel it is necessary to live in hope that the cause of the illness can be discovered; that some treatment can be found, if not to cure, then at least to relieve the symptoms, and that help will be available in caring for those who have to be hospitalized.

I keep hoping that a scientist will come up with something that would help. But without the money from Congress and donations from people who care, my family, my relatives, and other people with Huntington's will be as lost and helpless and confused as we were twenty years ago.

There were times when depression set in that I wanted to end it all. At the time I thought I didn't want to be treated the way they did my mom. No one even came to her funeral. But now I have learned to live each day as it comes, knowing what I know now about my illness. Each day I live in hope—that there will be hope for me and my kids; they took them because of my illness. I now can live in hope for a cure with the work of the Commission.

Budget

Budget Development for the National Plan

The criteria used in developing the budget were that dollar amounts would be allocated only for programs designed specifically for Huntington's disease patients and families and those with related disorders. Generic programs with widespread benefits are not included, with the exception of the Small Grants Program which is to promote research in the neurosciences in general.

Table 1
Total Budget Request

Budget Categories	FY 1979	FY 1980	FY 1981
A. Research	$8,900,000	$9,968,000	$11,164,160
B. Special Research Activities	360,000	298,000	333,760
C. Information Exchange	310,000	201,600	225,792
D. Centers Without Walls	2,080,000	2,329,600	2,609,152
E. Care	3,650,000	4,088,000	4,578,560
F. Education	400,000	350,000	392,000
G. Implementation of Huntington's Disease National Plan	75,000	84,000	94,080
TOTALS:	**$15,775,000**	**$17,319,200**	**$19,397,504**
Total Three-Year Budget Request: $52,491,704			

Table 2
A. Research

Recommendations	Responsible Agency	FY 1979	FY 1980	FY 1981
Research Grants related to Huntington's Disease (a) and (b)	NINCDS	$5,000,000	$5,600,000	$6,272,000
	NIGMS	1,400,000	1,568,000	1,756,160
	NIMH	1,100,000	1,232,000	1,379,840
	VA	400,000	448,000	501,760
Small Grants Program	NINCDS	1,000,000	1,120,000	1,254,400
TOTALS:		**$8,900,000**	**$9,968,000**	**$11,164,160**

(a) Figures based on the estimated cost of $100,000 per grant.
(b) Figures represent new money requests.

Table 3 B. Special Research Activities				
Recommendation	**Responsible Agency**	**FY 1979**	**FY 1980**	**FY 1981**
National Tissue Bank	NINCDS	$175,000	$150,000	$168,000
Venezuelan Study	NINCDS	150,000	143,000	160,160
Patient Rosters (a)	NINCDS	35,000	5,000	5,600
TOTALS:		**$360,000**	**$298,000**	**$333,760**

(a) Includes development and maintenance costs.

Table 4 C. Research Information Exchange				
Recommendation	**Responsible Agency**	**FY 1979**	**FY 1980**	**FY 1981**
Interdisciplinary Workshops (a)	NINCDS	$150,000	$168,000	$188,160
Neurogenetic Newsletter (b)	NINCDS	30,000	33,600	37,632
International Symposium on Huntington's Disease (c)	NINCDS	80,000	———	———
NIMH Symposium: Psychological Response to Neurologic Conditions (d)	NIMH	50,000	———	———
TOTALS:		**$310,000**	**$201,600**	**$225,792**

(a) Includes 24 meetings a year at an average cost of $4,300 per meeting. Program administration, production, printing, and distribution of workshop reports, approximately $46,800 annually, comprise the balance.

(b) Includes development, publication, and distribution costs of $1.50 per copy for 2-6 page quarterly newsletter for an estimated 5,000 investigators and other professionals.

(c) To organize and conduct a 3-day symposium consisting of 10 panels of 5 people each; includes travel, honorarium, per diem and other costs.

(d) To organize and conduct a 2-day symposium consisting of 6 panels of 5 people each; includes travel, honorarium, per diem and other costs.

Table 5 D. Centers Without Walls (Cost of One Center Only)				
Recommendation	**Responsible Agency**	**FY 1979**	**FY 1980**	**FY 1981**
Personnel (a)	NINCDS	$140,000	$156,800	$175,616
Core Support for Clinical Facility (b)		400,000	448,000	501,760
Research (c)		500,000	560,000	627,200
TOTALS:		**$1,040,000**	**$1,164,800**	**$1,304,576**

(a) Includes a center director, clinical staff, secretarial support staff, and consultants.
(b) Includes supplies, equipment, travel and evaluation.
(c) Supports research at the clinical facility or research associated with the facility.

Table 6 E. Care				
Recommendation	**Responsible Agency**	**FY 1979**	**FY 1980**	**FY 1981**
Respite Care Pilot Program (a)	NINCDS	$1,000,000	$1,120,000	$1,254,400
Health Services Coordination and Development Program (b)	CDC/NINCDS	800,000	896,000	1,003,520
NIMH Pilot Program (c)	NIMH	350,000	392,000	439,040
Model Long-term Care Facility Pilot Program (d)	NINCDS	1,200,000	1,344,000	1,505,280
Work and Recreation Day Care Pilot Program (e)	NINCDS	300,000	336,000	376,320
TOTALS:		**$3,650,000**	**$4,088,000**	**$4,578,560**

(a) Includes 2 pilot programs; each program provides 60 days of respite care a year for approximately 50 people.
(b) Includes 2 community programs.
(c) Includes pilot programs in 5 Community Mental Health Centers.
(d) Includes pilot programs in 2 facilities.
(e) Includes 3 pilot programs.

Table 7 F. Education				
Recommendation	**Responsible Agency**	**FY 1979**	**FY 1980**	**FY 1981**
Development and Distribution of Educational Materials (a)	NINCDS	$400,000	$350,000	$392,000
TOTALS:		**$400,000**	**$350,000**	**$392,000**

(a) Includes the development and distribution of 4 films, 15-20 minutes in length with back-up brochures; creation, shipping and staffing of HD exhibit; development and distribution of radio/TV minute spots; writing of five papers for professional publications; development and distribution of a clinical care handbook.

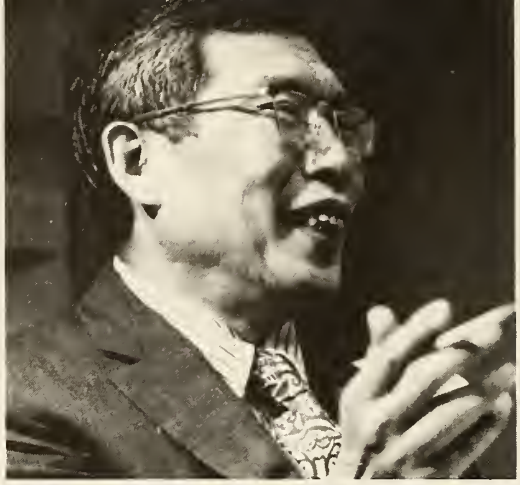
Table 8 G. Implementation				
Budget Category	**Responsible Agency**	**FY 1979**	**FY 1980**	**FY 1981**
Staffing of an NINCDS Branch for Huntington's Disease and related neurological disorders (a)	NINCDS	$75,000	$84,000	$94,080
TOTALS:		**$75,000**	**$84,000**	**$94,080**

(a) Includes Branch Chief, secretary, and clerk.

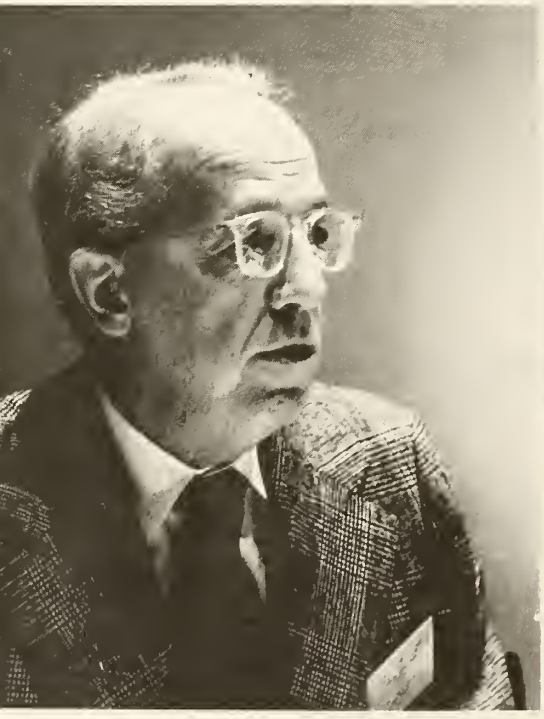
Recommendations: Implementing Agency and Legislative Requirements

Recommendation	Addressed to	Branch of Government or Private Agency	New Legislation Required
Research: Increased financial support for research in the neurosciences and genetics	Congress	NINCDS, NIGMS, NIA, NIMH, VA	None
Special Research Activities			
• Presymptomatic Detection	NINCDS	NINCDS	None
• Venezuela Project	Congress	NINCDS	None
• National Tissue Bank	Congress/ Secretary, HEW	DHEW	None
• Patient Rosters	NINCDS	Private or State Health Agencies/ NINCDS	None
• Interdisciplinary Workshops	NINCDS	Health Voluntary	None
• Neurogenetic Newsletter	NINCDS	NINCDS	None
• Conferences and Symposia	NINCDS/NIMH	NINCDS/NIMH	None
• Small Grants Program	NINCDS	NINCDS	None
• General Clinical Research Centers	Congress	DRR	None
• Increased funding for Research Training	Congress	NINCDS, NIGMS, NIMH, NIA	None
New Drug Development: an interdepartmental task force on development of new drugs for small populations afflicted by crippling diseases	President	Executive branch	None
Centers Without Walls	Congress/ Secretary, HEW	DHEW, NINCDS	None
Coordination of Health Services	Congress	NINCDS, CDC	None
Comprehensive Health Care for the Chronically Ill included in any program of National Health Insurance	President/ Congress/ Secretary, HEW	DHEW	Yes
Correct deficiencies in programs under the Social Security Act	Congress/ Secretary, HEW	DHEW, SSA, State Governments	Yes

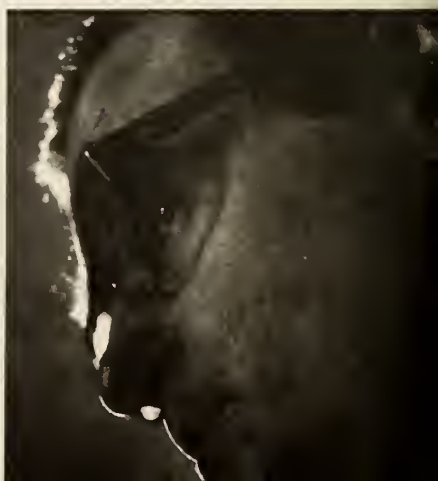
Recommendation	Addressed to	Branch of Government or Private Agency	New Legislation Required
Continuum of Care for Huntington's disease patients and others with chronic or disabling conditions	State Governments	State and local Health Systems Agencies, State Health Planning and Development Agencies	None
Pilot Programs: Long-term care; Respite Care; Work and Recreation Day Centers	Congress	NINCDS	None
New Health Care Personnel under CETA	DOL	DOL	None
Genetic Counseling: Training and Services	Congress	PHS	None
Demonstration Programs in Health Psychology	Congress/NIMH	NIMH	None
Employment opportunities for persons with Huntington's disease and other conditions	Secretary of Labor	DOL	None
Rehabilitation Services for persons with Huntington's disease and related disorders	Commissioner of RSA	DHEW/RSA/ State Vocational Rehabilitation Agencies	None
Education on Huntington's disease for families, health care and community personnel, general public	Congress	DHEW	None
Extension and full funding of the National Genetic Diseases Act	Congress	PHS	Extension of P.L. 94-278, Title IV
Unit in the Office of the Assistant Secretary for Health responsible for genetic services and research	Assistant Secretary for Health	DHEW	None
Implementation: a Branch for Huntington's Disease and Related Disorders in NINCDS	NINCDS	NINCDS	None







The Commission's Approach

The Commission for the Control of Huntington's Disease and Its Consequences was created by the Public Health Services Act, P.L. 94-63. The Commission held its first meeting in July 1976.

The Commissioners

The nine members, stipulated by law to include six professional and three lay persons, consisted of Marjorie Guthrie, New York, New York, Chairman; Doctor Milton Wexler, Los Angeles, California, Vice Chairman; Alice Pratt, Houston, Texas; Jennifer Jones Simon, Malibu, California; and Doctors Stanley Aronson, Providence, Rhode Island; Guy McKhann, Baltimore, Maryland; Ching Chun Li, Pittsburgh, Pennsylvania; Lee Schacht, Minneapolis, Minnesota; and Stanley Stellar, Livingston, New Jersey.

The professional disciplines represented on the Commission included neurology, neurosurgery, neuropathology, genetics, and psychoanalysis. The three lay commissioners were chosen for their special interest in public health or their personal involvement with Huntington's disease. The Executive Director, Doctor Nancy Wexler, is a clinical psychologist. The Deputy Director, Doctor Charles MacKay, is a specialist in biomedical ethics.

Public Hearings

The most compelling source of information for the Commission came from Huntington's disease patients and families. They wrote letters by the hundreds to the Commission office, and they spoke at public hearings held by the Commission in 11 cities throughout the country. These cities, Boston, New York,

Geographic Areas Corresponding to the Eleven Public Hearing Sites

Washington, Atlanta, Chicago, Ann Arbor, Wichita, Dallas, Denver, Los Angeles, and Seattle, were chosen as regional centers to represent all 50 states, and drew upon rural and urban populations in each area. The words of patients and family members, amplified by health care providers, public officials, and scientists were eloquent and often extraordinarily moving in describing the experience of living with Huntington's disease and the problems faced in finding adequate treatment and care. Quotes used throughout this report are taken from this testimony.

The Commission's major recommendations —for increased funding for basic research, for ways to develop new drugs, for financial aid, for improvements in the quality and quantity of care facilities and services—are a direct reflection of the needs expressed by these patients and families, as well as the considered advice of experts who counseled the Commission throughout the development of the Plan.

Interdisciplinary Work Groups

The Commission's principal technical source of guidance for scientific and clinical information came from 15 Commission work groups in the areas of biomedical research and social management. (See p. 100 for a list of work groups and members.) Members included Nobel laureates and other eminent scientists, leading clinicians and experts on Huntington's disease, and young scientists and clinical investigators exploring new areas of research and diagnostic technologies.

While the work groups were organized around specific disciplines in science or health care, membership was interdisciplinary. Often individuals with special knowledge such as architects, lawyers, educators, genetic counselors, and persons familiar with patient care were included. The work groups advised the Commission about the state of the art of research or management of Huntington's disease, made recommendations, and provided valuable data to aid the Commission in developing its program.

In some instances the work groups compiled the first systematic review of activities, or organized new data on research, care, and treatment of Huntington's disease, so that their reports constitute important additions to the literature. (See Vol. III for Work Group Reports.)

Position Papers/Scientific Meetings

Supplementing the information and recommendations from the work groups, the Commission used position papers prepared by professionals in such fields as the legal rights and problems of Huntington's disease patients, issues involved in new drug development, and ethical concerns in relation to the development or use of a presymptomatic test for Huntington's disease.

The Commission also invited interest and comments from clinical or research scientists by holding its meetings in conjunction with major scientific gatherings whenever possible. The advice and comments of the scientists attending these meetings were added to the ideas and recommendations the Commission solicited from world authorities.

Commission Studies

The Commission also cooperated with its sister Commission on epilepsy to explore areas of mutual interest. Federal programs relevant to the health or other service needs of their two constituencies were surveyed. The two Commissions also conducted a study to identify areas of common needs or interests for all patients with neurological, psychiatric, or degenerative conditions, preparatory to planning for combined action.

The Health Services Research Center of the University of California at Los Angeles surveyed Huntington's disease patients, families, and health care providers to assess the major medical, psychological, social, and financial needs that Huntington's disease produces. They sought to determine what barriers or gaps in the health care system impede access or availability of services. The work group on Behavioral Changes and Treatment compiled important personal and clinical data on 267 male and female Huntington's disease patients from across the country, the first such extensive data collection to be done.

The Health Voluntary Organizations

The Commission is deeply indebted to the Huntington's disease health voluntary organizations: the Committee to Combat Huntington's Disease, the Huntington's Chorea Foundation, the Hereditary Disease Foundation, and the National Huntington's Disease Asso-

ciation. These groups were an invaluable source of aid and advice during the course of the Commission's work. They helped to conduct surveys, organize public hearings, and encouraged the support of their membership. They did much to excite interest and inform the public about Huntington's disease and the work of the Commission. Above all, they were enthusiastic and appreciative of the opportunity for their members to be heard.

Over the scant decade of their existence the private health organizations have become the mainstay of support for patients and families. They have reached out into the community to locate new patients and provide much needed information. They have acted as liaisons trying to match family needs with existing community services. They have educated the professional as well as the lay public about the ravages of Huntington's disease. And they have been responsible for generating a groundswell of scientific interest in a hitherto little-known disease. They have supported valuable research on Huntington's disease and enabled scientists new to the area to begin work. They have organized scientific workshops and conferences on Huntington's disease which have sparked new approaches to the problem.

These organizations will continue to function as the first line of comfort and aid for patients and families and as an important stimulus and support for research. With the development of the National Plan their role can expand to include close cooperation with local, state, and Federal officials. They can help in the development of new care facilities such as respite programs or Work and Recreation Day Centers. They can be invaluable in developing programs in cooperation with Centers Without Walls, CMHCs, or in working with officials in the coordination of health services. They can assist in the generation of educational materials and help in the training of new health care personnel through their hard-won experience of the illness. The assistance of the voluntaries in research will grow as more researchers enter the field. Programs of tissue collection which the voluntaries have already initiated will enable scientists to conduct vital research—work which cannot be done without the cooperation of families.

The Commission wishes to express its gratitude to these dedicated volunteers. They and their members have eased the work of the Commission. Their continued commitment to the treatment, care, and eventual cure of Huntington's disease is creating an ever larger circle of friends and allies eager to help. That force for change will be the means by which the National Plan of the Commission will be accomplished.

I have lived with Huntington's for more than 40 years. First I watched my husband's mother die from the disease, which was not diagnosed as such. Next, my husband. I watched my husband die by inches leaving me with three children to raise. Financial burden was tremendous after he lost his job, insurance cancelled, etc. The emotional problems involved in trying to keep the family together and make a living were great with three children "at risk." At 17 years my son became a victim, just after his father's death. After 10 heartbreaking years watching him slowly die, I buried him just four years ago at age 27. All this time seeking any medical help or advice I could get—but nothing really helping, nothing concrete to go on. My husband died in a Masonic Hospital. I was forced to put my son in a state institution.

Now, four years later my daughter, age 34, is definitely showing symptoms of this dread disease. I can see it in her coordination and change in personality. She is still teaching and I pray she will be able to continue her work, because her husband walked out leaving her with a nine year old daughter to support and, of course, who now is "at risk."

Glossary

ACETYLCHOLINE: A compound in the body which affects muscle action, inhibits heart activity and influences digestion and dilation of the blood vessels.

ACHONDROPLASIA: An inherited autosomal dominant disease causing congenital dwarfism through defective conversion of cartilage into bone.

AGE OF ONSET: Age at which symptoms of a disease first appear.

ALLELE: One of several alternative forms of a gene occupying a particular position on a chromosome.

ALZHEIMER'S DISEASE: Progressive dementia caused by severe damage to the cells in the brain. This fatal disease usually occurs in persons under 50 years of age.

AMNIOCENTESIS: Extraction and analysis of a sample of the watery fluid in which the embryo is suspended in the uterus.

AMYOTROPHIC LATERAL SCLEROSIS (ALS): A chronic, progressive disease caused by degeneration of the nerve cells in the spinal cord and marked by progressive muscle weakness and atrophy.

ANIMAL MODEL: An animal that exhibits the same patterns of a certain disease as humans and can therefore be used in tests for treatment of the disease and other research.

ANTIBODY: A protein produced by the body in response to a foreign substance, with the specific capacity to create immunity to and neutralize that substance.

ANTIGEN: A substance capable of provoking the body to make an antibody against it.

ANTIGEN-ANTIBODY COMPLEX: A bonding of an antigen and its specific antibody.

ATAXIA: Loss or lack of muscular coordination.

ATROPHY: The wasting away of tissues, organs, or the entire body.

AUTOSOMAL INHERITANCE: A form of transmission of genetic characteristics not related to sex as a result of the gene concerned being carried on the autosome.

AUTOSOMES: Chromosomes other than sex chromosomes.

BASAL GANGLIA: Areas of brain tissue located at the base of the cerebrum, primarily influencing motor control.

BIOLOGICAL CLOCK: The biological mechanism responsible for the periodicity of certain behavior in living organisms.

CARRIER: An individual harboring a specific disease who, though often not exhibiting the symptoms, may transmit the disease to others. In genetics, a carrier means an individual whose chromosomes contain the gene for a specific genetic condition.

CELLULAR BIOLOGY: The study of the structure and processes of living cells.

CEREBELLAR ATAXIA: Loss of muscular coordination as a result of disease in the cerebellum—sometimes hereditary.

CEREBELLUM: The part of the brain responsible for regulation and coordination of complex voluntary muscular movement.

CEREBRAL CORTEX: The outer layer of gray tissue of the brain largely responsible for higher mental processes, such as thinking, perception and memory.

CEREBRAL PALSY: Difficulty in control of the voluntary muscles caused by a prenatal brain defect or by brain injury during birth.

CEREBROSPINAL FLUID (CSF): The serum-like fluid that bathes portions of the brain and the cavity of the spinal cord.

CEREBRUM: The largest part of the brain, consisting of two halves or hemispheres.

CHOREA: A nervous disorder characterized by irregular, involuntary movements of the muscles of the body.

CHROMOSOMES: The thread-like bodies in the cell nucleus that are responsible for carrying hereditary characteristics from one generation to the next. Each chromosome is made up of a number of genes.

CHRONIC DISEASE: A disease of slow progression and long duration.

COMPUTERIZED AXIAL TOMOGRAPHY (CAT): Computerized technique for making X-ray pictures of a predetermined object, usually the central part of the body (head or trunk), by blurring out the outline of the other objects in the focal field.

COMPUTERIZED POSITRON TOMOGRAPHY: Computerized technique for making X-rays of parts of the brain by injecting or inhaling radioactive isotopes.

COOLEY'S ANEMIA: Severe recessively inherited disorder of the red blood cells. Onset occurs in infancy or early childhood.

CREUTZFELDT-JAKOB DISEASE: This rare, progressive disease of the brain and spinal cord is characterized by dementia and weakening and stiffening of the limbs. It is probably caused by one of the slow viruses and is found in the middle-aged and elderly.

DEMENTIA: Mental deterioration due either to organic or psychological factors.

DIABETES (MELLITUS): A disease that impairs the ability of the body to use sugar and causes the abnormal appearance of sugar in the urine.

DEOXYRIBONUCLEIC ACID (DNA): A nucleic acid found in the nucleus of cells, and functioning in the transference of genetic characteristics.

DOMINANT: A genetic trait which masks the contrasting recessive trait in an individual.

DOPAMINE: A chemical substance essential to normal nerve cell activity in the brain.

DOWN'S SYNDROME: A syndrome of mental retardation caused by chromosomal abnormalities. Characteristics include retarded growth, short, flattened skull, slanting eyes and thickened tongue. Also known as "Mongolism."

DYSKINESIA: Difficulty in performing voluntary movements.

DYSLEXIA: Impairment of the ability to read with understanding, due to central nervous system malfunction.

DYSTONIA (MUSCULORUM DEFORMANS): An affliction marked by muscular contractions producing distortions of the spine and hips. There are three types of dystonia: the autosomal recessively inherited form, the autosomal dominantly inherited form, and acquired dystonia.

ELECTROENCEPHALOGRAM (EEG): A graphic record of the electrical activity of the brain.

ELECTROPHORESIS: The influence of an electric field on the movement of particles toward a positive or negative pole. Used as a technique in biochemical analysis.

ELECTROPHYSIOLOGY: The study of electrical phenomena that are associated with physiologic processes.

ENCEPHALITIS: Inflammation of the brain.

ENCEPHALOPATHY: Any disease of the brain.

ENDEMIC DISEASE: A disease continually present in a community or among a group of people.

ENDOCRINOLOGY: The branch of medicine dealing with the internal secretions of the body.

ENZYMES: The proteins in the body functioning as biochemical catalysts in metabolic processes.

EPIDEMIOLOGY: The study of the frequency and distribution of a disease in a community.

EPILEPSY: A chronic brain disorder of various causes characterized by recurrent seizures due to excessive discharge of cerebral neurons.

FAMILIAL HYPERCHOLESTEROLEMIA: An autosomal dominant hereditary disorder characterized by the presence of abnormally large quantities of cholesterol in the blood, and leading to heart disease.

FRIEDREICH'S ATAXIA: A loss of the power of muscular coordination, caused by a hereditary nervous system disorder.

GAMMA-AMINOBUTYRIC ACID (GABA): A chemical substance in the central nervous system which inhibits nerve impulses.

GENE: The functional unit in the cell chromosome responsible for carrying hereditary characteristics from one generation to the next.

GENETIC COUNSELING: Giving of information to an individual regarding the hereditary nature of an illness and assistance in decisions stemming from this information.

GILLES DE LA TOURETTE'S DISEASE: A disease beginning in childhood and characterized by motor incoordination resulting in bizarre tics and unusual speech patterns such as involuntary repetition and use of vulgar and obscene words.

GLUCOSE METABOLISM: The sum of the physical and chemical processes that blood sugar undergoes in the tissues of an organism.

GLUTAMIC ACID DECARBOXYLASE (GAD): A chemical substance found in the brain which is involved in the formation of gamma-aminobutyric acid (GABA).

HALLUCINOGENIC DRUG: A drug that produces hallucinations.

HEMIBALLISM: Violent spasmodic movements involving one side of the body.

HEREDITARY TREMOR: An inherited disease characterized by trembling, shaking, or shivering.

HUNTINGTON'S DISEASE: An autosomal dominant disorder, with onset usually in middle age, characterized by abnormal involuntary movements (chorea), progressive intellectual impairment (dementia), and a spectrum of psychiatric disturbances. The disease is progressive and terminal over a period of 10 to 20 years.

HYPOTHALAMUS: The part of the brain that regulates many basic body functions, such as body temperature and appetite.

IMMUNITY: Resistance to a substance perceived as a foreign invader, such as a virus.

INCIDENCE: The number of new cases of a specific disease occurring during a certain period.

INHIBITORY TRANSMITTERS: Chemical agents which have a restraining effect on the firing of nerve impulses.

KAINIC ACID: A chemical substance used in experiments on animals that destroys the same cells in the animal brains as are destroyed in human brains by Huntington's disease.

KURU: A progressive fatal disease of the brain found in certain Melanesian tribes in New Guinea, assumed to be of viral origin.

L-DOPA: Drug used for the treatment of Parkinson's disease.

LINKAGE: The association of genes in inheritance, due to the fact that they are on the same chromosome.

LITHIUM CARBONATE: A drug used in the treatment of the manic phases of manic-depressive states.

LYMPHOCYTE SENSITIZATION EXPERIMENTS: Studies of the white blood cells to determine their degree of sensitization against a disease.

MANIC-DEPRESSIVE PSYCHOSIS: Mental disease where periods of manic excitation alternate with melancholic depression.

MOLECULAR BIOLOGY: The study of the composition and activities of the molecules making up living matter.

MULTIPLE SCLEROSIS: A chronic, degenerative neurologic disease found chiefly among young adults and caused by hardening of the tissues of the brain and/or of the spinal cord.

MUSCULAR DYSTROPHY: Inherited progressive wasting of the voluntary muscles of the body.

NARCOLEPSY: A condition characterized by sudden and uncontrollable attacks of deep sleep, occurring at irregular intervals.

NEUROANATOMY: The science of the structure of the nervous system.

NEUROBIOLOGY: The study of the functions of the brain and the nervous system.

NEUROLOGICAL DISORDERS: Organic diseases of the nervous system.

NEURONS: Nerve cells.

NEUROPATHOLOGY: The science that deals with the causes and development of diseases of the nervous system.

NEUROPHARMACOLOGY: The study of drugs that produce an effect on nervous tissue.

NEUROPHYSIOLOGY: The study of the functions and processes of the nervous system.

NEUROPSYCHIATRIC DISORDERS: Organic and functional diseases of the nervous system.

NEUROSIS: Psychological or behavioral disorder often characterized by anxiety.

NEUROTRANSMITTER: A chemical substance influencing transmission of nerve impulses.

NOSOLOGY: The systematic classification of diseases.

PARKINSON'S DISEASE: A progressive nervous disease causing muscular tremors, rigidity of movement, and peculiarity of gait, posture, and facial expression.

PENETRANCE: The frequency, usually expressed as a percentage, with which a gene produces its specific effect in its carriers in a population.

PHENOTYPE: A category or group to which an individual may be assigned on the basis of characteristics resulting from both its heredity and its environment.

PICK'S DISEASE: Rare, presenile dementia causing intellectual deficits, speech difficulties, and emotional instability.

PITUITARY GLAND: A small gland attached to the base of the brain whose secretions control the other glands and influence growth, metabolism and maturation.

PLATELETS: Small, irregular disk-shaped elements in the blood vital to the clotting process.

POLYGENES: Interacting genes, each producing a small part of the total effect of the group.

POLYPOSIS: An autosomal dominant disorder characterized by the presence of polyps in the colon which usually become malignant.

PRESENILE DEMENTIA: Dementia appearing in patients under 60, characterized by intellectual deficits and emotional instability due to various causes.

PRESYMPTOMATIC TEST: Tests used for the detection of the existence of a disease before any symptoms have appeared.

PREVALENCE RATE: The rate at which a certain disease exists in the population.

PROSTHETIC AIDS: A fabricated substitute for a missing part of the body, such as a limb, tooth, eye, or heart valve.

PROTOTYPE: A perfect example of a particular type.

RADIOACTIVE ISOTOPES (RADIOISOTOPES): Unstable nuclear substances that decay to a stable state by emitting characteristic radiation

RECESSIVE: A type of genetic trait, the effect of which may be masked by the presence of a dominant trait.

ROBOT SMOKER: A mechanical prosthetic aid designed to allow patients with muscular disorders to manipulate cigarettes.

SCHIZOPHRENIA: A severe mental disorder resulting in withdrawal from reality and emotional and intellectual disturbances.

SCRAPIE: A disease of sheep and goats causing various nervous symptoms and usually leading to death.

SENILE DEMENTIA: An organic brain disease associated with aging and marked by progressive mental deterioration and emotional instability.

SHINGLES: A viral infection of certain sensory nerves characterized by clusters of blisters following the path of the affected nerve.

SICKLE CELL ANEMIA: Recessively inherited disorder characterized by abnormalities in the oxygen-carrying red blood cells, causing them to become sickle-shaped.

SLOW VIRUS ENCEPHALOPATHY: A disease of the brain, following a long, unremitting course.

SPASM: An involuntary muscular contraction.

SYDENHAM'S CHOREA: A toxic or infectious nervous disease occurring in young persons, causing involuntary muscular movements of the face, neck and limbs.

TARDIVE DYSKINESIA: A drug-induced disorder manifesting excessive muscular activity that persists after drug withdrawal and appears to be irreversible.

TAY-SACHS DISEASE: A rare, recessive hereditary disease causing mental retardation, paralysis and death in early childhood.

THALASSEMIA: A recessively inherited chronic disorder of the oxygen-carrying material of the blood ranging from early detectable abnormalities to severe and fatal anemia.

TISSUE CULTURE: A specimen of living tissue taken from the body and cultivated in an appropriate medium.

VIROLOGY: The study of viruses and viral diseases.

Abbreviations/Acronyms

CAT: Computerized Axial Tomography
CDC: Center for Disease Control
CETA: Comprehensive Employment and Training Act
CMHC: Community Mental Health Center
CSF: Cerebral Spinal Fluid
DHEW: Department of Health, Education, and Welfare
DHUD: Department of Housing and Urban Development
DOL: Department of Labor
DOT: Department of Transportation
DRR: Division of Research Resources, National Institutes of Health
EEG: Electroencephalogram
FDA: Food and Drug Administration
GABA: Gamma-aminobutyric Acid
GCRC: General Clinical Research Centers
HCFA: Health Care Financing Administration
HSA: Health Services Administration
IND: Investigational New Drug
NIA: National Institute on Aging
NIGMS: National Institute of General Medical Sciences
NIH: National Institutes of Health
NIMH: National Institute of Mental Health
NINCDS: National Institute of Neurological and Communicative Disorders and Stroke
NRSA: National Research Service Awards
PHS: Public Health Service
PSRO: Professional Standards Review Organization
RSA: Rehabilitation Services Administration
SSA: Social Security Administration
SSI: Supplemental Security Income
TAPS: Training and Placement Service
VA: Veterans Administration

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Montefiore Hospital, the Bronx

Members:

Edward Bird, M.D.
Addenbrooke's Hospital
Cambridge, England

Murray Bornstein, M.D.
Albert Einstein College of Medicine

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Uniformed Services University of the Health Sciences

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Veterans Administration Hospital
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Montefiore Hospital, the Bronx

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Children's Hospital Medical Center
Boston

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New York State Psychiatric Institute

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New York State Psychiatric Hospital

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College of Physicians and Surgeons
Columbia University

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Albert Einstein College of Medicine

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National Institute of Neurological and Communicative Disorders and Stroke

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College of Physicians and Surgeons
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Office of Independent Living
Department of Housing
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Massachusetts General Hospital

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Boston

Arthur L. Benton, Ph.D.
University Hospital
Iowa City, Iowa

Nelson Butters, Ph.D.
Veterans Administration Hospital
Boston

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National Institute of Mental Health

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The Johns Hopkins University Hospital

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Center for Disease Control

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Medical Center

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Mayo Clinic

Manfred Muenter, Ph.D.
Mayo Clinic

Ntinos Myrianthopoulos, Ph.D.
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Neuropsychiatric Institute, Los Angeles

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University of Rochester

Kavi Berg, M.D.
University of Oslo

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Indiana University Medical Center

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Hereditary Disease Foundation

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Addenbrooke's Hospital
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Hospital Universitario
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Communicative Disorders and Stroke

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Office of Science and Technology Policy
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Richard A. Prindle, M.D.
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Raymond Roos, M.D.
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Albert Einstein College of Medicine

Heber Villalobos, M.D.
Hospital Universitario
Maracaibo, Venezuela

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9701 Wilshire Blvd.
Beverly Hills, CA 90212

Telephone: (213) 274-5443

National Huntington's Disease Association
1441 Broadway, Suite 501
New York, NY 10018

Telephone: (212) 966-4320

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